

**Favourable Results from Predictive Testing in Huntington's Disease: An
Exploration of the Long—term Impact on Close Relationships.**

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Declaration

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I declare that the present thesis is my own work and that the work of which it is a record has been done by myself. No work contained in the thesis has been submitted in application for a degree in any other institution or university. Any personal data has been processed in accordance with the provisions of the Data Protection Act 1998. All quotations have been distinguished by quotation marks and the sources of information specifically acknowledged.

Dianne Beastall 2011

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Abstract

Background: Huntington's disease (HD) is a chronic neurodegenerative condition caused by a genetic mutation. HD is incurable and affects a person's cognitive, behavioural, emotional and motor functioning. Symptoms typically develop around 30 – 45 years old with life expectancy approximately 15-20 years from onset. Children of an affected parent are at 50 per cent risk of inheriting the disease, and those individuals who inherit the abnormal gene will eventually be affected by HD. The HD gene was discovered in 1993 which resulted in a direct genetic test which could confirm the absence or presence of HD in at-risk individuals. There is a large amount of research into the psychological consequences of predictive testing, with more interest in those individuals who receive unfavourable test results. Less is known about those who receive favourable results and it has been suggested that those individuals do not always experience uncomplicated relief.

Research Aim: The study aimed to explore how receiving a favourable result from predictive testing for HD can impact on close relationships in the long-term.

Methodology: A constructivist grounded theory approach was used to meet the aim of the study, generating data through in-depth interviews with 10 people who received favourable results from predictive testing for HD at least five years previously. The interviews were transcribed verbatim and analysed line-by-line using a series of coding procedures.

Findings: The findings suggest that when people find out they are at risk of HD they face a journey into the unknown and have to try to manage the uncertainty related to living at risk. Making the decision to be tested ends this uncertainty and once people receive a favourable result from predictive testing for HD, they go through a process of trying to distance themselves from HD. Those who have close family members (siblings in particular) with HD or who are at risk of HD can find this process more challenging. Having a sense of duty to family members with HD explains some of the difficulties faced by the participants.

and Background to the Study

Conclusions: The findings of this study could help raise awareness of the long-term issues and support needs affecting individuals who receive favourable results from genetic testing. Implications of the findings for clinical practice are discussed, and recommendations for research are made.

Chapter 1

Introduction

“For a few months we were reeling, as this illness has so many elements to it that we had to absorb and come to terms with – your parent being terminally ill, yourself being at risk, your children being at risk, too; what to tell and not to tell the children: it was as if our lives were unravelling. Then my dad went on to die of the disease. We saw at firsthand how it can dismantle someone. Anyway life was about to change completely” (Sulaiman, 2007, p.24).

1.0 Background to the Study

The above quote captures the emotional distress which a woman experienced when she discovered that her father had Huntington’s disease (HD) and then realised the implications this had for her and the rest of the family. She was subsequently diagnosed with HD and wrote a book (Sulaiman, 2007) about her individual family members’ experiences of living with HD. In terms of the present study, this book acted as a starting point in exploring the impact which HD has on individuals and families. The decision to read the book was prompted by early discussions with a Consultant Clinical Geneticist about her experiences of working in the area of HD and pre-symptomatic predictive genetic testing for HD. Of particular interest were the dilemmas faced by at-risk individuals when deciding whether to go for predictive genetic testing and the impact this and the test results have on the individual and their family members. This chapter’s structure will follow the psychological journey of at-risk individuals from before predictive testing to a number of years after receiving the test result. Research examining the pre-test stage and the decision-making process of at-risk individuals will be reviewed followed by an examination of studies which assess the psychological

functioning of at-risk individuals at various stages after genetic testing. The chapter will then review research which investigates the wider impact of predictive genetic testing for HD and considers the position of spouses, partners and the family of at-risk individuals. The chapter will conclude by outlining the research aim for the current study.

1.1 Terminology

For the purposes of this thesis the terms “unfavourable” and “favourable” are used to refer to positive (presence of HD gene) and negative (absence of HD gene) test results respectively. The term “carrier” is used to describe at-risk individuals who have received unfavourable results from predictive testing, and the term “non-carrier” refers to those individuals who received favourable genetic test results (Duisterhof *et al.* 2001).

1.2 Huntington’s Disease

In order to set the study in context it is necessary at this point to provide some basic information about HD. HD is defined as: “*a neurodegenerative disease characterised by involuntary movements (chorea), progressive dementia, and affective disturbances (for example aggression, paranoia)*” (Decruyenaere *et al.* 1996).

Dawson *et al.* (2004) report that 30 per cent of individuals with HD have symptoms consistent with a diagnosis of depression, and suicide is common in the HD population. This is possibly explained by the fact that HD is incurable which makes it all the more devastating for affected individuals and their families. Rawlins (2010) reported that HD

prevalence studies have been carried out in a number of regions of the United Kingdom and that the overall average is 6-7 per 100,000 of the population. Rawlins explained that this rate is an underestimation of the true prevalence of HD given that the Huntington's Disease Association (HDA) currently provides care for 6702 people with HD in England and Wales, which means the prevalence must be 12.4 per 100,000 of the population. This rate is also an underestimation because the HDA does not provide services in all areas of England and Wales and there will be people with HD who do not get referred to the HDA (Rawlins, 2010). Possible reasons for the difficulties in establishing the prevalence of HD include the stigma of the disease which causes families to hide the presence of the disease from others including health professionals (Rawlins, 2010). Although HD is relatively rare the impact of the disorder can be widespread in terms of the effect on carers and family members (Quarrell, 2008).

There is a 50 per cent risk of children inheriting HD if a parent is affected (Timman *et al.* 2004). It is not possible to carry the gene and not develop HD (Dawson *et al.* 2004). If the gene is passed on from parent to child then the child will definitely develop the disease, at some stage usually during mid-adulthood. Signs typically develop around 30 – 45 years old with a survival rate of approximately 15–20 years from onset (Keenan *et al.* 2007). As an at-risk person increases in age the likelihood of developing HD gradually declines. However, due to the unpredictable age of onset, at-risk individuals who decide against going for testing can never be sure if they have been spared of the disease (Evers-Kiebooms & Decruyenaere, 1998). The early stages of the disease can include symptoms such as uncontrollable jerky movements, clumsiness, concentration difficulties, memory problems, fluctuating mood and occasionally aggressive behaviour

(Huntington's Disease Association, 2008). These symptoms can threaten relationships particularly for individuals who are unaware that family members are at risk of HD (Huntington's Disease Association, 2008). In addition, a lack of awareness of HD being in the family history, coupled with the late presentation of HD symptoms can result in reproductive decisions being made with potential costs for subsequent offspring (Chapman, 2002). Individuals with HD often have life changes enforced upon them as the symptoms of HD progress, for example leaving their job, having less social contact with friends, and losing their mobility and independence (Dawson *et al.* 2004). Some individuals with HD experience a more rapid decline in their motor and cognitive functioning whereas others can continue in employment, and maintain positive relationships for a comparatively longer time period despite their impairments (Kessler, 1993b). During the more advanced stages of HD residential care is often required in order to meet the support needs of the individual; however, there are limited specialist facilities for people with HD (Dawson *et al.* 2004).

1.3 Predictive Genetic Testing and Huntington's Disease

Before the introduction of predictive genetic testing for HD, individuals at risk of the disease had no option but to make important life choices such as entering into long-term relationships and having children whilst being unaware of their genetic status (Evers-Kiebooms & Decruyenaere, 1998). In 1983 it was discovered that the gene for HD was located on chromosome four (Guisella *et al.* 1983). A few years later pre-symptomatic predictive genetic testing became possible through linkage analysis which involves using gene markers that are closely related and requires multiple family members to participate in testing (Harper *et al.* 2000). The test made it possible for at-risk

individuals to establish with a high degree of certainty whether the HD gene had been passed on to them, prior to the onset of symptoms (Codori, Hanson & Brandt, 1994). Evers-Kiebooms *et al.* (2002) acknowledged the importance of the identification of the HD gene in the 1980s for medical history. They also highlighted the implications this development had for professionals and families in terms of being faced with difficult decisions about whether to be tested or not. Smith *et al.* (2002) pointed out that the accessibility of pre-symptomatic testing provides individuals at risk with the option of obtaining information which can have a significant impact on their lives and future plans. Codori *et al.* (1997) listed the possible advantages of receiving an unfavourable genetic test result as removing feelings of uncertainty, having the opportunity not to transmit the gene and enabling careful planning for the future. Despite these potential benefits, individuals who receive unfavourable results from predictive testing have to face a life where deteriorating functioning and premature death are inevitable (Dudok de Wit *et al.* 1998).

A further advance occurred when The Huntington's Disease Collaborative Research Group (1993) discovered an expanded, unstable trinucleotide repeat on the HD chromosome. They anticipated that identifying this mutation in individuals at risk of HD would be a landmark development in predictive testing. They stated that complex linkage analyses would no longer be required and the new predictive test would be an option for at-risk individuals who have no living relatives affected by HD (The Huntington's Disease Collaborative Research Group, 1993). Harper *et al.* (2000) reported that it is uncommon for a single mutation to be responsible for a genetic disorder as is the case with HD. Pre-symptomatic predictive genetic testing involving

mutation analysis is currently available as a clinical service in NHS settings in the United Kingdom and in many countries throughout the world (Harper *et al.* 2000). The genetic test for HD is approximately 100 per cent sensitive and specific for identifying the gene mutation (Dawson, *et al.* 2004).

Harper *et al.* (2000) collected data on all pre-symptomatic genetic tests for HD in the UK for the ten year period from 1987-1997. They found that 2937 tests had been undertaken during this time, with 2502 tests involving mutation analysis which has been possible since 1993. Interestingly, it is estimated that approximately 18% of at-risk individuals present for predictive testing for HD (Harper *et al.* 2000). There were significantly more females (58%) than males participating in testing and it has been proposed that this might be because of reproductive decisions and being more prepared to address difficult choices and their implications (Harper *et al.* 2000). The average age at testing was 36.2 years old and there were very few individuals less than 20 years old. There were large numbers of individuals aged over 50 who participated in testing, and it was suggested that older individuals might request testing in order to help their children who might be of an age where family planning issues are relevant (Harper *et al.* 2000). Abnormal results were received by 41.4% of individuals and by 29.4% of those aged 60 and above. Harper *et al.* (2000) comment that this highlights the huge variability in age of onset of HD symptoms and state that older individuals might minimise the risk of HD and give little thought to the consequences of an abnormal result.

Individuals can be at 25 per cent risk of HD if they have an at-risk parent who has not been tested and is either alive or has died without displaying symptoms of the disease

(Benjamin & Lashwood, 2000). If individuals with a 25 per cent risk decide to go for predictive testing then the result has implications for parents or siblings who may not wish to have this knowledge. During the first two years of direct predictive testing 85 tests were carried out on individuals at 25 per cent risk across 23 genetic departments in the UK, and 54 (63.4%) parents were living at the time of the test (Benjamin & Lashwood, 2000). Approximately two-thirds of parents were offered genetic counselling either directly or through the child; however, there was little interest with only 12 parents agreeing to counselling. Benjamin and Lashwood (2000) state that all genetic testing centres in the UK advise the inclusion of both the 25 per cent risk individual and parent if possible in genetic counselling and promote an open approach between family members. They highlight the importance of counselling and psychological input particularly in cases where complicated decisions are required, as is the case with individuals at 25 per cent risk.

1.4 Genetic Counselling

Due to the concerns by health professionals about the psychological effects of pre-symptomatic genetic testing for HD a counselling protocol was developed (Smith *et al.* 2002). For those individuals who express an interest in genetic testing the protocol recommends they attend three pre-test counselling appointments which involve receiving information about HD and the genetic testing process in order to help inform their decision (Smith *et al.* 2002). The pre-test counselling sessions also encourage and support the test candidate to anticipate what their life would be like if they receive an unfavourable result, a favourable result or if they decide not to proceed with predictive testing (Evers-Kiebooms, *et al.* 2002). Williams *et al.* (2000) state that a time period of

at least two months is necessary in order to meet the requirements of the gene test protocol and for the test results to be communicated to test candidates if testing goes ahead. Test candidates usually receive their test results in person at a meeting with a Consultant Clinical Geneticist or a genetic counsellor.

Since the discovery of the HD gene mutation in 1993, it is possible for test candidates to be told whether or not they have the faulty gene. The presence of the HD gene indicates that the person will develop HD at some point in their life but the test is unable to reveal when (Huntington's Disease Association, 2011). There are four different types of results which at-risk people can receive. The faulty gene contains the genetic code, CAG (cytosine-adenine-guanine) which is repeated many times. If the person has less than 27 repeats then the result is unequivocally normal and the person will not develop HD. A result which reveals a repeat size of between 27 and 35 is normal but there is a small risk that the repeat may expand in future generations. If there are between 36 and 39 repeats then the result is abnormal; however there is a chance the person may not develop HD until late in life and in some cases not at all. A repeat size of above 40 is unequivocally abnormal and the person will develop HD (Huntington's Disease Association, 2011). Quarrell (2008) reports that there is variation in how laboratories report the test result with some including the specific repeat size and others only including the broad category result (e.g. normal, normal but in the 27-35 range, abnormal in the 36-39 range or unequivocally abnormal). Quarrell (2008) explains that in his own clinical practice he chooses not to disclose the repeat size to the individual unless the specific size is requested. He states that if a person wants this information then he explores the reasons with them before giving the repeat size. Once the

predictive test result has been revealed emotional support is offered both in the short and long-term (Evers-Kiebooms & Decruyenaere, 1998).

Duisterhof *et al.* (2001) report that prior to the advent of pre-symptomatic testing for HD there was only one study which investigated the psychological well-being of individuals at risk for HD. This lack of research interest was surprising to them as they proposed that it could be reasonable to assume that psychological issues would be prevalent in those at risk who have grown up in families significantly affected by HD. There was an increase in psychological research when clinicians and researchers became more aware of the negative impact which pre-symptomatic genetic testing could potentially have on people at risk of HD (Duisterhof *et al.* 2001).

1.5 Decisions about Predictive Testing

The introduction of predictive testing has presented at-risk individuals with the option of finding out if they have inherited the mutated HD gene. A number of studies have been carried out to establish the reasons for and against going for predictive testing. Evers-Kiebooms *et al.* (1989) explored the motivating factors and attitudes of at-risk individuals and their partners prior to participating in predictive testing. Interestingly, the results suggested an eagerness of at-risk individuals (66%) and their partners to participate in genetic testing. However, approximately one third of those individuals had no intention in going for the test immediately. Evers-Kiebooms *et al.* (1989) expressed their surprise at this but noted that it was consistent with the low-uptake of testing at other genetic centres despite a great deal of initial interest in taking the test. Kessler (1994) suggested that although the intense and lengthy nature of the genetic testing

process is important, it could also explain why there has been a poorer uptake in testing than expected and it is possibly only the most motivated individuals that participate in testing. Individuals at risk who were in favour of the predictive test gave reasons such as planning for the future and to have certainty. For those who already had children they felt it was important to be able to tell their children about their risk of HD (Evers-Kiebooms *et al.* 1989). Reasons for not wanting to take the test included finding it difficult to live with an adverse test result, preferring a life of uncertainty and there being no cure or treatment for HD (Evers-Kiebooms *et al.* 1989).

Similar research methods have been used by other studies into decision-making regarding pre-symptomatic testing for HD. Tibben *et al.* (1993a) used an attitude questionnaire, in which the majority of the questions were multiple-choice, to examine the pretest attitudes and expectations of at-risk individuals and their partners. They found that family planning was the main reason for participants taking the genetic test followed by reducing uncertainty. Partners of those at-risk chose planning for the future and family planning as the most significant reasons for testing. The reasons selected for not taking the test included fear of the negative consequences of an unfavourable result, developing a preoccupation with searching for symptoms and adopting the sickness role before the onset of symptoms.

Tibben *et al.* (1993a) proposed that individuals who apply for the test have an expectation that they will not experience an emotional reaction to either test outcome. This is consistent with Codori *et al.*'s (1994) findings that most people who had gone through genetic testing for HD did not anticipate problems associated with the test result

and even those who thought they would experience a difficult emotional reaction were confident of their coping ability. In contrast, Codori *et al.* (1994) found that at-risk people who decided against testing were more likely to anticipate psychological distress in relation to the test result. Therefore they suggested that people who go for predictive testing are self-selected for a psychologically healthy reaction to testing. Binedell *et al.* (1998a), however, advise caution in negatively labelling people who do not request testing, as being more psychologically vulnerable with poorer coping abilities. Instead they propose that deciding not to have the test should be perceived as a legitimate choice and may indicate that the at-risk person is more psychologically able to tolerate the uncertainty surrounding their genetic status.

Although these studies provided an interesting insight into factors which influence the decision to take part in predictive testing, the use of questions with mainly pre-coded answer categories limited the opportunity for participants to describe their experiences in detail. This method of data collection perhaps also masked the complexity of the decision-making process. Binedell *et al.* (1998a; 1998b) used both quantitative and qualitative methods in their research into the differences between people at risk who requested predictive testing and those at risk who had decided not to be tested. They used verbatim responses to structured questions as opposed to an in-depth exploration of experiences which could have provided more rich information.

Smith *et al.* (2002) used qualitative methods to illuminate the complex psychological processes which are involved when deciding whether or not to go for predictive genetic testing for HD. All of the participants in the study were women and they talked about

how hard it was to cope with their genetic status. The interviews also revealed that the 50 per cent risk is misinterpreted by some participants and that lay beliefs about genetic transmission were evident, for example that the gene can miss a generation or only females in a family can get HD. These misinterpretations and beliefs then resulted in participants either perceiving an increased or decreased risk of HD. The findings revealed that the decision to take the test or not was not part of an informed decision making process. Instead the decision was often made before making contact with the genetic clinic, family history and experiences influenced the decision and there was a sense of not wanting to take responsibility for making the decision (Smith *et al.* 2002).

Quaid *et al.*'s (2008) qualitative study involved exploring the everyday experiences of people living at risk of HD who have chosen not to undergo predictive genetic testing. The core theme which emerged from the data was 'living at risk', of which concealing risk and preserving hope were important main themes. The findings revealed that participants at risk of HD were very careful at concealing their genetic status and only divulged this information to people they trusted in situations where they felt in control (Quaid *et al.* 2008). In terms of preserving hope, the participants felt able to tolerate the uncertainty of living with the risk of HD by maintaining hope that they did not have the disease or that as they get older their risk decreases. Quaid *et al.* (2008) commented on the way in which the participants managed to find an incredible balance between risk and hope in their everyday lives, and advised healthcare professionals to be cautious about suggesting that having knowledge of one's HD risk is preferable to not knowing.

1.5.1 Reproductive Decisions

Planning to have children has been reported as one of the main reasons to contemplate predictive testing (Tibben, 2007). Decruyenaere *et al.* (1996) found that predictive testing had a significant impact on reproductive decisions one year post-test disclosure. Approximately two thirds of carriers who had planned to have a family decided not to have children or take part in prenatal testing, whereas the majority of non-carriers went ahead with their plans to have children. Decruyenaere *et al.* (1996) highlight the complex dilemma which carriers face if they want children but do not want to pass on the mutated gene given that each pregnancy carries a 50 per cent risk of termination. Evers-Kiebooms *et al.* (2002) list the options for carriers of the HD mutation who want to start a family: deciding not to have children, being prepared to risk the chance of the child having the HD mutation, using prenatal testing/diagnosis, artificial insemination with donor sperm, IVF with donor eggs, pre-implantation genetic diagnosis (PGD) and adoption.

A study involving seven genetic test centres across six European countries gathered data on reproductive decision-making after predictive testing for HD in both carriers and non-carriers (Evers-Kiebooms *et al.* 2002). It was found that there was a sizeable impact of predictive test results on family planning in that twice as many non-carriers went on to have at least one pregnancy compared to carriers. Prenatal diagnosis was carried out in approximately 66 per cent of carriers and one couple opted for PGD which led to a successful pregnancy and birth. Predictive testing appeared to have a greater impact on reproductive decisions in the subgroup of individuals who stated that family planning was their main reason for participating in predictive testing (Evers-

Kiebooms *et al.* 2002). In this group 39 per cent of carriers became pregnant compared to 69 per cent of non-carriers, which suggests that there were no pregnancies after testing in the majority of carriers within the period of three to seven years post-testing (Evers-Kiebooms *et al.* 2002). Evers-Kiebooms *et al.* (2002) acknowledge that family planning as a reason for predictive testing could have different meanings for different test candidates, such as not wanting to transmit the mutated gene to their offspring or not wanting their children to grow up with a parent who is affected by HD.

In a similar study carried out in Australia, Richards and Rea (2005) did not find a significant difference between the number of post-test pregnancies in carriers and non-carriers. They proposed that reproductive decisions of at-risk individuals in Australia might be less influenced by predictive testing than their counterparts in Europe. However, Richards and Rea (2005) suggest that the non-significant difference could be attributed to the loss of data to follow-up in 50 participants who were mainly non-carriers, younger in age and had no children before testing. Richards and Rea (2005) reported that is in keeping with similar studies there was very little interest in prenatal testing and other fertility procedures by carriers and individuals at risk of HD. The reasons for the low uptake were not explored with participants in this study; however Richards and Rea (2005) speculated that carriers had reservations about terminating a pregnancy for a disease that does not develop until adulthood. They provided anecdotal evidence which described the distress of two couples in the study who had undergone previous terminations after receiving high-risk prenatal results and then decided not to participate in prenatal testing for future pregnancies.

Low uptake of prenatal testing was also reported in Canada in the period 1986-1991 (Adam *et al.*, 1993). There were 47 pregnancies out of the 425 individuals who participated in predictive testing and only 14 (30%) couples requested prenatal testing. Nine couples (19%) received a favourable result from predictive testing which meant they did not require prenatal testing, and 24 (51%) couples stated that they were not interesting in prenatal testing. Seven couples ended up withdrawing from the prenatal testing which meant only seven took part in the procedure (18%) (Adam *et al.* 1993). The most common reasons for not participating in prenatal testing was the hope that a cure for HD would be found during their children's lifetime and the wish to have a child overshadowed the risk that the child could develop HD (Adam *et al.* 1993). Participants in this study had to choose their reasons for not requesting prenatal testing from a list and it is possible that this resulted in important information being missed.

The complex nature of the reproductive decision-making is frequently highlighted in the research; however, this is masked by many studies employing questionnaire methods to gain insight into participants' reasoning. Although quantitative research investigating the impact of predictive testing on reproductive decisions is useful in terms of identifying trends, frequencies and correlations, a clearer understanding of the reasoning behind participants' reproductive decisions could be obtained through qualitative research.

A recent study by Decruyenaere *et al.* (2007) has responded to this gap in the research by using mixed methods to identify factors which influence reproductive decision-making in carrier couples post predictive testing. Decruyenaere *et al.* (2007) decided to

use a longer follow-up period and interviewed participants approximately five years post-test result. Analysis of the interviews revealed that reproductive decision-making for carrier couples was complex and involved a variety of issues. Having the option of prenatal diagnosis and PGD was perceived as a way to reduce distress, but at the same time the carrier couples questioned the ethics of having an abortion when the child could live a healthy life for the first 30 or 40 years (Decruyenaere *et al.* 2007). Other factors which influenced the decision to have children included concerns about offspring growing up with an affected parent and observing their declining functioning, and finding it psychologically difficult to have a child knowing that they are at risk of HD. It was found that the carrier's own experiences of growing up with an affected parent could influence their own decision to have children particularly if they had a difficult relationship with their affected parent and questioned their suitability to care for children (Decruyenaere *et al.* 2007). Doubts about a cure for HD being found in the foreseeable future contributed to some carrier couples decision not to have children. Decruyenaere *et al.* (2007) highlight the importance of pre- and post-test genetic counselling for exploring issues around family planning given the complicated and emotional nature of reproductive decision-making in carriers of the mutated gene.

Klitzman *et al.* (2007) also conducted a qualitative study which provided rich and detailed information about the dilemmas which carriers, non-carriers and untested at-risk individuals face when considering having children. The analysis revealed that individuals battled to work through numerous questions relating to reproduction such as whether they should have any children, get pregnant and participate in prenatal testing, pre-implantation genetic diagnosis or have a termination (Klitzman *et al.* 2007). A

model was created which illustrates the numerous decisions faced by individuals and how they were often stuck between their personal wishes against their perceived responsibility towards their partner, family, current and future children and the wider society (Klitzman *et al.* 2007). It also emerged that over time some individuals in the study changed their reasoning and viewpoints with each pregnancy, for example taking a risk with the first pregnancy and then having prenatal testing for the second pregnancy (Klitzman *et al.* 2007).

It is clear that although there are some commonalities between the dilemmas faced by carriers and at-risk persons the reasoning is highly individual and should be explored thoroughly in genetic counselling. Having considered the research on the complexities of decision-making in relation to predictive testing and family planning for individuals at risk of HD the next section will now focus on the psychological consequences of predictive testing for HD.

1.6 Psychological Effects of Predictive Testing for HD

In a review of the research on the psychological impact of predictive testing for HD, Meiser and Dunn (2000) reported that the majority of studies in this area suggest that there is a significant difference between carriers and non-carriers in levels of psychological distress in the short-term but not in the long-term. Outcome measures used in the studies included depression, anxiety, general well-being and hopelessness. From the studies reviewed it emerged that psychological adjustment to the test result appeared to depend more on adjustment before predictive testing than the impact of the actual test result. Van't Spijker and ten Kroode (1997) carried out a review of the

psychological aspects of genetic counselling for HD and found that in a number of studies, carriers reported short-term emotional responses after receiving their non-favourable result. A variety of emotions were described such as numbness, sadness, depression, anxiety and anger, with levels of depression and anxiety generally decreasing to normal levels within one year post test result.

At the time of Van't Spijker and ten Kroode's (1997) review there had been no reports of long-term psychological responses to predictive testing for HD. Other important findings were that non-carriers and partners of test applicants also experienced emotional reactions to the test result such as depression, numbness and guilt. Broadstock *et al.* (2000) conducted a systematic review into the psychological consequences of predictive testing for HD and other hereditary conditions and found that at-risk people taking part in testing do not experience negative psychological reactions up to three years post-test-result. This finding could be explained by evidence that people who go for testing are emotionally stronger and self-selected for a favourable response to the result, and most of the studies had a follow-up period of 12 months or less (Broadstock, *et al.* 2000).

Wiggins *et al.* (1992) proposed that predictive testing for HD has possible advantages for the emotional wellbeing of both carriers and non-carriers. In their study they found that non-carriers demonstrated a considerable improvement in psychological wellbeing at 10 days, 6 months and 12 months post-test result. The carriers did not show severe psychological reactions to testing that was first anticipated. The authors suggested that having knowledge of favourable and unfavourable genetic statuses helps to decrease

uncertainty and allows people to plan for the future. They acknowledged that the participants in their study were generally well-educated and middle-aged which is possibly not representative of all individuals who take part in predictive testing. The participants were also part of an intense testing program which included genetic counselling and psychological support and it is questionable whether the results of the study would generalise to people who are part of a less rigorous protocol.

Tibben *et al.* (1994) also found that carriers did not demonstrate severe psychological reactions to the test result. When the test result was revealed carriers reported an increase in pessimistic expectations; however, these returned to baseline levels at 6 months post-test result. There was a decrease in intrusive thoughts about HD for both carriers and non-carriers during the 6 month period following the test result. The authors suggest that the results indicate feelings of relief from the at-risk status for both groups but also suggest that carriers may minimise or deny the effect of the test result. This denial was also observed by Tibben *et al.* (1993b) in a 6 month follow-up interview with carriers in that most of them rated their current life circumstances as being good. As with similar studies in this area, the generalisability of the results is questionable due to the higher educational levels of the participants and the sustained psychological support they received. In addition, the 6 month follow-up period is relatively short and it may take longer than this for adjustment problems and psychological distress to emerge (Tibben *et al.* 1994).

Lawson *et al.* (1996) found that in the year following the predictive test result 14.8% (n=135) of participants experienced an adverse event (e.g. relationship breakdown,

increase in substance misuse, psychiatric hospitalisation, and suicide attempt or plan). There were no significant differences in terms of the occurrence or type of adverse events between carriers and non-carriers but the timing of the adverse events differed significantly. Carriers tended to experience an adverse event within the first 10 days following the test result whereas non-carriers did not have an adverse event until at least 6 months post-test result. Participants with a previous history of depression were more likely to experience an adverse event in the year after testing. Codori *et al.* (1997) reported that those individuals who had more difficulty adjusting to their test result in the year following testing were more likely to have received a positive test result, were married, had no children or were nearer to their anticipated age of onset of HD.

Following up participants in predictive testing for up to one year post-test result is perhaps not a long enough time period for psychological problems to emerge. It may be much later that both carriers and non-carriers experience distress when the carriers start to show symptoms and the non-carriers are having to care for a parent or siblings who are in the advanced stages of the disease. However, there are far fewer studies exploring the long-term psychological consequences of predictive testing and therefore there is a lack of knowledge in this area (Almqvist *et al.* 2003). Tibben *et al.* (1997) found that there was no significant difference between carriers and non-carriers in changes in distress and hopelessness from baseline to three years after the test result. The authors were surprised by this finding given the prognosis for carriers of the HD gene and conversely the greater life opportunities for non-carriers. They speculated that the high rate of drop-outs (33%) at the three year follow-up could have accounted for this unexpected result.

In contrast to this, Almqvist *et al.* (2003) reported a significant decrease in psychological distress in both the carriers and non-carriers at two years and five years post-test. Adverse events (suicide, suicide attempts and psychiatric hospital admission) were experienced by 14 participants (6.9%) and were more likely to occur in the first year following the test result with significantly more adverse events happening for those who received an increased risk test result (Almqvist, *et al.* 2003). Interestingly, the participants who received a decreased risk test result experienced the most severe incidents such as suicide attempts (Almqvist, *et al.* 2003). Although the adverse events are happening for relatively small numbers of individuals it is still important that health professionals know that suicide and psychiatric illness can occur amongst this group. In a more recent study, Larsson *et al.* (2006) found that both carriers and non-carriers demonstrated high suicidal ideation prior to predictive testing, and in the two years following the result carriers' levels of depression and suicidal thoughts increased compared to non-carriers. Although the dropout rate was very small in this longitudinal study, the overall sample size was also small which limits the generalisability of the findings.

In their study into the psychological effects of predictive testing 7-10 years after the result, Timman *et al.* (2004a) propose that research which has revealed minimal negative effects may have underestimated the real impact. In their study it was found that carriers who dropped out of the study had significantly higher pretest scores on hopelessness, intrusion, avoidance and lower levels of wellbeing than carriers who remained in the study for the follow-up period. Another important finding was that for

carriers, levels of hopelessness were higher than baseline at one week after the test but then it reduced significantly to its lowest point 18 months after the test. Thereafter hopelessness levels started to rise again and were higher than pre-test levels at 7-10 years after the test result. This possibly could be explained by carriers reaching the age of onset of HD and starting to notice symptoms. Timman *et al.* (2004a) recommend that longitudinal studies into the impact of predictive testing should use longer follow-up periods than a few years in order to better understand the psychological impact of the test result. They also highlight the problem of researchers not providing information on the problem of participants dropping out of longitudinal studies, and advise them to include the data of individual who have dropped out in order to gain a more detailed picture of the psychological functioning of all individuals participating in testing.

Timman *et al.*'s (2004b) interest in the statistical quality of longitudinal studies into the psychological effects of predictive testing prompted them to carry out a systematic review in this area. They selected 40 studies for the review but most of them used inadequate statistical methods. The problems included not reporting on the dropout rate and failing to provide information on missing values and the features of dropouts. Timman *et al.* (2004b) argue that if more suitable statistical techniques had been used then a large number of studies could have produced more accurate results.

1.6.1 The Psychological Impact of Receiving Favourable Results

It may be assumed that receiving a favourable result from genetic testing for HD would be a positive experience with feelings of relief and happiness (Huggins *et al.* 1992). Whilst this may be true of some at-risk individuals it is apparent from the literature that

the situation is less straightforward. Individuals who receive favourable results experience a greater amount of psychological problems than was anticipated by most health professionals (Van't Spijker and ten Kroode, 1997).

A recent study found that out of 64 non-carriers, 27 per cent reported that they struggled to cope with their favourable genetic test result and a significant amount of non-carriers were depressed (24%) at an average of 3.7 years post test-result (Gargiulo *et al.* 2009). Three non-carriers had attempted suicide in the five years following their favourable test result suggesting it takes considerable time to adjust to being 'not at risk' and highlighting the need for long-term psychological support in both carriers and non-carriers (Gargiulo *et al.* 2009). Similarly, Tibben *et al.* (1997) found that in terms of levels of hopelessness, non-carriers future outlook had not changed over a period of 3 years. They proposed that this could be because the favourable result had not impacted on their life as expected and they realised it did not provide an escape from HD in their families.

Kessler (1994) highlighted the need for health professionals working in genetics to re-evaluate their assumptions about what at-risk individuals deem as a good or bad result. He explained that a favourable result might not be viewed positively by some at-risk individuals if it is contrary to their expectations and conscious/unconscious hopes which can then lead to low mood and anxiety. Kessler (1994) provided case examples from his own experience to illustrate how initial relief on receiving a favourable result can then change to mood problems and adjustment difficulties, with individuals unsure how

to live life without the threat of HD, and also have to face the unexpected negative reactions of other family members to their news.

Similarly Huggins *et al.* (1992) found that at least 10 per cent of individuals with a decreased risk result in their genetic testing program experienced psychological problems in adjusting to their test result. Huggins *et al.* (1992) admitted that this reaction had not been anticipated by them and they use a series of case reports to highlight the circumstances surrounding the adverse responses. Common themes that emerged across the cases included individuals possessing unrealistic expectations that a favourable result would improve their life, being convinced that they would receive an increased risk result, making irreversible decisions that stemmed from the belief they had the HD gene, for example having a vasectomy and overspending resulting in huge debts, and for some individuals survivor guilt was a problem (Huggins, *et al.* 1992). In one of the case reports it was the spouse who struggled to adjust to the decreased risk result of his wife as he had a strong intuition that his wife would develop HD and had planned to care for her during his retirement. Huggins *et al.* (1992) recommend that genetic counsellors engage in a thorough assessment during pre-test counselling in order to identify factors which make individuals who receive a decreased risk more vulnerable to psychological difficulties. Tibben *et al.* (1993a) found that prior to going for pre-symptomatic testing, test applicants more frequently than their partners anticipated that a decreased risk result would result in an improvement in their ability to plan for their future. Only small numbers of applicants expected improvements in their quality of life and their relationships.

The anecdotal reports used by Kessler (1994) and case examples presented by Huggins *et al.* (1992) have raised important issues about the psychological functioning of individuals receiving a favourable result. However, their observations are based on clinical experience instead of research. Despite the suggestions in the literature that individuals who receive favourable results might struggle to adjust to their new genetic status there is a lack of research in this area. Only one study could be found which focuses on individuals who have received favourable results from predictive testing for HD. Williams *et al.* (2000a) state that there is a lack of knowledge about the consequences of finding out that one has not inherited the gene mutation for HD. They conducted a qualitative study which explored the psychological impact and adjustment processes of receiving favourable results from predictive testing for HD and pallid-ponto-nigral degeneration (PPND). Participants were interviewed initially before receiving their test result and then again at one and six months after their test result. Williams' *et al.* (2000a) described a process of redefinition which participants went through after receiving their favourable result. The redefinition process related to views about self, position in the family and role in society.

Williams *et al.* (2000a) explained that the participants had predicted how they would react to an unfavourable result but had not considered the consequences of discovering that they did not have the gene mutation. Participants discussed the difficulties the favourable result had on family relationships in terms of not sharing the common bond of being at risk and needing to choose carefully the family members to which they disclosed their status (Williams *et al.* 2000a). At six months post-test result the redefinition process was still occurring but participants were less focused on themselves

and were starting to shift their attention to the future (Williams *et al.* 2000a). This study is the first study to explore in-depth the experiences of at-risk individuals who receive a favourable predictive test result for HD. The authors called for further research which examines the redefinition process within a family context.

Van Riper (2005) used the cases of two families to illustrate the complex issues which can arise when family members go for genetic testing for HD and for breast and ovarian cancer. The families were selected from a larger ongoing study into the ethical issues relating to families going for genetic testing. Analysis of the case-studies revealed that although genetic testing was mainly a positive experience for the family at risk of HD, they experienced unexpected negative consequences (Van Riper, 2005). The two sisters who received favourable results had convinced themselves that they would develop HD and had made future plans with this in mind. Telling family members about the favourable result caused difficulties for the sisters and on the day one sister found out her test result she had to attend the funeral of her uncle who had HD.

1.6.2 Impact of Predictive Testing on Relationships

Initially, research on the psychological impact of predictive testing tended to focus on at-risk individuals; however, there has been an increasing interest in their spouses and partners (Richards, 2004). Quaid and Wesson (1995) was the first study to investigate the impact of predictive testing on intimate relationships. They found that spouses had significantly higher levels of depression than their at-risk partners before testing, highlighting the importance of including spouses in genetic counselling. Of the 25 couples who participated in genetic counselling, six decided not to proceed with

predictive testing and it was found that at baseline they had significantly more psychological distress and marital problems than the couples who participated in testing. It was suggested that couples with stronger partnerships engaged in predictive testing as they perhaps believed that their relationship could cope with the pressure of receiving the test result.

Quaid and Wesson (1995) reported that couples who found out that they were at high-risk of HD had higher levels of distress after testing than those who received low-risk results. However, it was highlighted that although there was an increase in distress, all scores were still within the non-clinical range. The authors acknowledged their small sample size and contend that the participants benefited from intense counselling and support which other test candidates might not have access to thus questioning the representativeness of their sample. It is possible that the couples who withdrew either before or after testing had higher levels of distress and coping difficulties (Quaid & Wesson, 1995).

Decruyenaere *et al.* (2005) found that carriers' partners were as distressed as the carriers themselves at 5 years post-test and in some cases felt worse, more fearful and shocked, less competent, more angry and passive than carriers. The authors emphasised the need for health professionals to be aware of the distress of carriers' partners after predictive testing and to offer them the opportunity to discuss their concerns.

Kessler (1993a) has found that in his experience spouses can be indifferent about their at-risk partner finding out if they have the HD gene but are generally supportive of their

partner's decision. Regardless of the result the spouse will possibly have to change their perception of their future relationship and potentially find new ways of coping. Kessler (1993a) used case reports to illustrate the impact which HD can have on the spouse. He raises the issue of secrecy and explains how some at-risk individuals who are aware of their family history of HD keep this private due to concerns that they will be rejected by their spouse. Kessler (1993a) reports that there appears to be a higher likelihood of marital/relationship separation in couples where the at-risk individual has kept HD a secret until symptoms develop.

Although the case reports in Kessler (1993a) are useful in presenting real-life examples of the impact of HD on at-risk individuals and their partners, they do not provide an in-depth account from a personal perspective of what it is like to be an at-risk individual or partner in this context. Richards (2004) carried out the first study which explored the experiences of individuals at risk of HD and their partners regarding living with the risk of HD, predictive testing, and how this has impacted on their relationship. Using a qualitative approach Richards (2004) conducted interviews with 14 couples of which six had undergone predictive testing (three carriers and three non-carriers), five had not been tested, and three were displaying symptoms of HD. The findings indicated that for the participants in the study it is the psychological distress associated with living at risk of HD that is the root of the relationship problems and not the impact of the specific test result. Richards (2004) highlight the importance of health professionals being mindful of the unique nature of each couple's relationship.

1.6.3 A Family Perspective

Tibben (2007) describes the devastating impact which HD can have on a family system through having to cope with a variety of losses and changes such as physical deterioration and personality changes in the affected person, loss of the original family system and death of family members. He reports that in addition they are affected by issues such as shame, secrecy and social isolation. Writing from personal experience, Hayes (1992) emphasises the need for health professionals to view HD from a family perspective and not focus solely on the affected individual. She explains that a HD diagnosis has an impact on the entire family and members cope in a variety of different ways.

Preselection is a coping strategy used by some families with HD (Kessler, 1988). It is defined as: *“the singling out, in advance, of an asymptomatic relative to become eventually the ‘affected’ individual”* (Kessler, 1988, p.618). Preselection is not performed at a conscious level by family members, and the preselected individual is usually doesn’t realise what his or her psychological function is within the family (Kessler, 1988). Preselecting an at-risk relative helps the family to believe that there has been a decrease in the many unknowns associated with HD thus giving them a sense of control and containing their anxiety. Kessler (1988) explains that the preselected individual is often a child and it usually first occurs when the child’s parent has started to display signs of HD. If a child has the same name as an affected parent then this can increase his or her likelihood of being preselected, and often behavioural characteristics shared by the child and affected parent are pointed out which reinforces the illusion, for example “dad walked that way” (Kessler, 1988). The important issue about preselection

is that it can have a hugely detrimental effect on the child's functioning in that the family tend to divert their attention and resources on to the other children as they think that they have a future without HD. Kessler (1988) states that preselected children and adolescents are at risk of low self-esteem and depression.

The notion of preselection is an interesting concept which is worth bearing in mind when working with families at risk of HD. However, the evidence appears to be based on clinical experience as opposed to research studies. Van Riper (2005) states that there is a lack of understanding about the experience of genetic testing within a family context and emphasises the importance of focusing on the family perspective. Some researchers in the area of predictive testing for HD have incorporated theoretical frameworks into their studies to highlight and explain the impact of testing on the whole family (Sobel & Cowan, 2000a; Dudokdewit *et al.* 2002).

1.7 Theoretical Framework

The family life cycle (Carter and McGoldrick, 1989) is a useful theoretical framework through which to understand how predictive testing for HD can affect families. Using the family life cycle framework, Brouwer-Dudokdewit *et al.* (2002) presented 6 cases to illustrate the difficulties families with HD face when moving from one life stage to another. They proposed that HD causes significant disruption to the "normal" transitions within the life cycle and suggested that the actual test result causes less disruption than the families altered future expectations and opportunities. Tibben (2007) also uses a life-cycle perspective to highlight how HD can affect family relationships at different stages.

A description of Carter and McGoldrick's (1989) family life cycle stages is presented in Table 1. Carter and McGoldrick (1989) state that there is sufficient evidence to suggest that family stresses, which more commonly happen at transition points in the life cycle, often cause interruptions to the life cycle and create emotional problems. They explained that stress and anxiety within a family can occur horizontally and vertically. Vertical stress is passed down from previous generations such as attitudes, secrets and expectations. Horizontal stressors impact on the family as they progress through life, adjusting to the transitions within the family life cycle. These refer to both predictable developmental life stressors and unpredictable life events. Chronic illness is classified as an unpredictable life stressor and this could include HD but it is to some extent more predictable than other diseases. For example, if a person has had predictive testing they may know from a young age that they are going to develop the disease at some point and they also know that there is no cure and it is likely they will die approximately 15 years after the symptoms begin. On the other hand HD is unpredictable in that people do not know when the symptoms will start and if they decide not to go for testing or do not even know that they are at risk of HD then the illness is more of an unpredictable stressor. Huntington's disease could also be a vertical stressor in terms of it being kept a secret by some families or conversely knowing that the disease is genetic and can be passed down through generations can cause a great deal of anxiety for individuals and families.

Carter and McGoldrick (1989) propose that when the horizontal and vertical stressors intersect it can cause a significant increase in anxiety levels within the family system.

For example, if a young couple are expecting their first child and during the pregnancy find out the expectant mother is at risk of HD, then after the birth they have to cope with the usual stresses associated with expanding a family system but also cope with the knowledge that she and her child are at risk of the disease thus significantly increasing anxiety levels. In addition to horizontal and vertical stressors, Carter and McGoldrick (1989) propose a third type of stress and that is the social, financial, political and cultural stress associated with the current time period.

In terms of the stages of the family life cycle presented in table 1, HD can have a significant impact on all stages and transitions. There are different challenges and stresses specific to each stage, for example finding out about the risk of HD when a single young adult may cause significant anxiety about entering into intimate relationships and trying to plan ahead for the future. On the other hand finding out about the risk of HD once a person is married and has children is likely to cause a great deal of stress and worry about the future health of the person and their children. As the children approach mid-late adolescence and start getting into relationships it may prompt the at risk parent to go for predictive testing. The family life cycle can also be hugely affected if a family member starts showing symptoms of HD and there will be emotional demands placed on the entire family as the disease progresses. The family life cycle is a very useful and meaningful theoretical framework with which to make sense of the issues facing individuals and families living with HD.

Table 1: The Stages of the Family Life Cycle (Carter and McGoldrick, 1989).

Family Life Cycle Stage	Emotional Process of Transition: Key Principles	Second-Order Changes in Family Status Required to Proceed Developmentally
1. Leaving home: Single young adults	Accepting emotional and financial Responsibility for self	a. Differentiation of self in relation to family of origin. b. Development of intimate peer relationships c. Establishment of self re work and financial independence
2. The joining of Families through Marriage: The new couple	Commitment to new system	a. Formation of marital system. b. Realignment of relationships with extended families and friends to include spouse
3. Families with young children	Accepting new members into the system	a. Adjusting marital system to make space for child(ren) b. Joining in childrearing, financial and household tasks c. Realignment of relationships with extended family to include parenting and grandparenting roles
4. Families with adolescents	Increasing flexibility of family boundaries to include children's independence and grandparents' frailties	a. Shifting of parent child relationships to permit adolescent to move in and out of system b. Refocus on midlife marital and career issues c. Beginning shift toward joint caring for older generation
5. Launching children and moving on	Accepting a multitude of exits from and entries into the family system	a. Renegotiation of marital system as a dyad b. Development of adult to adult relationships between grown children and their parents c. Realignment of relationships to include in-laws and grandchildren d. Dealing with disabilities and death of parents (grandparents).
6. Families in later life	Accepting the shifting of generational roles	a. Maintaining own and/or couple functioning and interests in face of physiological decline; exploration of new familial and societal role options b. Support for a more central role of middle generation c. Making room in the system for the wisdom and experience of the elderly supporting the older generation without overfunctioning for them d. Dealing with loss of spouse, siblings and other peers and preparation for own death. Life review and integration.

1.8 Limitations of the Literature

Most studies into the psychological consequences of predictive testing for HD have used standardised psychological outcome measures to assess a variety of psychological factors at pre-test (baseline) and compared them at various time periods post-test. Most of the consequences assessed in the studies are related to emotion and there is a lack of cognitive and behavioural outcome measures (Broadstock *et al.* 2000). Furthermore, the measures that most studies have used are for general purposes and are not validated for use with people who have participated in predictive testing for HD which limits the generalisability of the findings (Duisterhof *et al.* 2001). Another limitation of the literature is the evidence that most people who proceed with predictive genetic testing are self-selected for a favourable response to testing. They usually have higher levels of education than the general population and are more often female. This means that the research findings from studies into the psychological consequences of predictive testing may not apply to people at risk of HD in the general population (Meiser & Dunn, 2000). Also most studies involve relatively small numbers of participants who are part of rigorous counselling and research protocols which questions the generalisability of the results to people who are not part of such programs.

1.9 Rationale and Aim of Present Study

Although the difficulties faced by people who have received favourable results from genetic testing for HD have been identified in the research, there are few studies which have explored these difficulties in-depth from the perspective of the person who received the result. As a consequence, there have been calls for more qualitative research in the area of predictive testing for HD (Binedell *et al.* 1998; Kessler, 1997;

Lawson *et al.* 1996; Richards, 2004; Tibben *et al.* 1997). The aim of the present study was to explore the experience of receiving a favourable result from predictive genetic testing for HD and how this has impacted on close relationships in the long-term. The study will focus on individuals who received a favourable result from testing at least five years ago as there is a lack of research on the long-term consequences of predictive testing (Almqvist *et al.* 2003).

Reports in the literature suggest that for some individuals it takes a considerable length of time to adjust to no longer being at risk of HD. The reason for interviewing people at least five years after their test result is that people at this later stage might have siblings who have the HD gene and have started displaying symptoms, or they might have to care for a parent or sibling who could be in the advanced stages of the disease. In addition, individuals will have been living without the risk of HD for at least five years allowing them to reflect on whether their lives are as they would expect them to be given they do not have HD.

1.10 Chapter Summary

The decision to research the psychological impact of predictive genetic testing for HD stemmed from discussions with a clinical geneticist and through reading about the personal experiences of a family living with HD. Having limited prior knowledge of this area it was important as a starting point to gain an understanding of the genetics of HD, the disease symptoms and progression and the genetic testing process. As the quote at the beginning of this chapter illustrates, HD is clearly a devastating illness with wide-ranging implications for affected individuals and their families. This is why it is

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1.10 Chapter Summary

The decision to research the psychological impact of predictive genetic testing for HD stemmed from discussions with a clinical geneticist and through reading about the personal experiences of a family living with HD. Having limited prior knowledge of this area it was important as a starting point to gain an understanding of the genetics of HD, the disease symptoms and progression and the genetic testing process. As the quote at the beginning of this chapter illustrates, HD is clearly a devastating illness with wide-ranging implications for affected individuals and their families. This is why it is

useful to use a theoretical framework such as the family life cycle to help make sense of the issues in the area of HD and predictive testing. Although the introduction of predictive testing has provided at-risk individuals with the opportunity to lessen their future uncertainty it has also presented them with hugely complex issues relating to self and family. Both quantitative and qualitative research studies have helped to increase our understanding about the scale and complexity of decision-making about predictive testing, family planning issues and also the psychological consequences of predictive testing for carriers and non-carriers. Despite evidence to suggest that people who receive favourable results from predictive testing for HD can experience psychological difficulties in adjusting to their result there is a lack of research which focuses on this population. The next chapter initially describes the main tenets and characteristics of grounded theory and justifies the selection of the approach for this study. The chapter then describes the research method through considering the ethical issues relating to the study, sampling issues, the data collection process, data analysis procedures and the steps taken to ensure the trustworthiness of the findings.

Chapter 2

Method

2.0 Introduction

The literature review presented in Chapter One revealed a lack of qualitative research exploring the experiences of people who receive a favourable result from predictive genetic testing for HD. The first half of this chapter briefly discusses qualitative research approaches, before describing in more detail the main characteristics of grounded theory, and justifies its use in the present study. The second half of the chapter outlines the ethical considerations for the study and the procedure adopted including participant recruitment and sampling issues, data generation and data analysis. The chapter ends with a description of how the trustworthiness of the findings were ensured.

2.1 Design

2.1.1 Qualitative Research

A qualitative design was adopted in order to meet the aim of the study which was to explore the experience of receiving a favourable result from predictive genetic testing for HD and how this has impacted on close relationships in the long-term. Qualitative research is defined as:

“an inquiry process of understanding based on distinct methodological traditions of inquiry that explore a social or human problem. The researcher builds a complex, holistic picture, analyzes words, reports detailed views of informants, and conducts the study in a natural setting” (Creswell, 1998 p.15).

Although the difficulties faced by people who have received favourable results from genetic testing for HD have been identified in the research, there are few studies which have explored these difficulties in-depth from the perspective of the person who received the result. Qualitative research was therefore considered appropriate for the present study due to its focus on the in-depth understanding of human experiences. Furthermore, Smith *et al.* (2002) state that qualitative research is beneficial for research areas that have complex issues as is the case with predictive genetic testing for HD.

There are a number of qualitative research approaches available to the researcher. For example phenomenology, discourse analysis and grounded theory are widely used in health research and although they are similar in nature they also have particular differences (Starks & Trinidad, 2007). A number of studies in the area of genetic testing for HD have used interpretative phenomenological analysis (IPA) as a research approach (e.g. Chapman, 2002; Chapman & Smith, 2002; Macleod, *et al.* 2002). IPA is concerned with the meaning of experiences for participants (Smith & Osborn, 2008) and whilst this is of interest in the present study, grounded theory was considered more suitable because it also focuses on process and explaining how participants respond to a particular experience (Morse, 2001). McAllister (2001) argues that there is a demand for evidence-based theories in genetic counselling and that researchers in this area are showing a growing interest in qualitative research methods. She highlights the contribution which grounded theory methodology can make to research about the psychosocial processes going on in relation to genetic counselling.

2.1.2 Grounded Theory

Grounded theory is the most commonly used qualitative research approach by researchers across a variety of disciplines (Bryant & Charmaz, 2010). It was developed by Glaser and Strauss in the 1960s who defined grounded theory as;

“the discovery of theory from data which fits empirical situations and is understandable to sociologists and layman alike and most important, it works - provides us with relevant predictions, explanations, interpretations and applications” (Glaser and Strauss, 1967, p. 1).

The publication, *The Discovery of Grounded Theory* (Glaser & Strauss, 1967) was written to satisfy the curiosity of people who had read Glaser and Strauss’ research on the awareness of dying and wanted to know more about the methods they had used (Glaser, 1992). Grounded theory’s logical and systematic approach comes from Glaser’s background in quantitative research whereas Strauss was influenced by symbolic interaction which is reflected in grounded theory’s focus on process, meaning and action (Charmaz, 2008). Glaser and Strauss (1967) argued that in sociological research there was a focus on verifying theory with very little interest in discovering or generating themes and hypotheses.

In grounded theory, researchers start with a general research aim or question and gradually develop a theory through analysing the research findings (Charmaz, 2008). Grounded theory is inductive in that the theory develops after data collection commences. However, it is also deductive in terms of analysing data and then deciding where or who to sample next (Glaser, 1978). Glaser and Strauss (1967) highlight the importance of the underlying process involved in developing theory which is the simultaneous collection, coding and analysis of data. They advise that researchers

should engage in all three tasks together as often as they can. Glaser and Strauss (1967) use the term, theoretical sampling to describe this process and define it as, “*the process of data collection for generating theory whereby the analyst jointly collects, codes and analyses his data and decides what data to collect next and where to find them, in order to develop his theory as it emerges*” (Glaser & Strauss, 1967, p.45).

Researchers stop collecting data when they have reached the point of saturation. This means that nothing new is coming out of the data and where gathering more data would probably not develop the theory or explanation much further (Strauss & Corbin, 1998). Strauss and Corbin (1998) also state that in reality, researchers often stop collecting data before saturation has naturally occurred, due to limits on their time or financial resources.

2.1.3 Development of Grounded Theory

Since grounded theory was first developed in the 1960's there have been a number of developments including a long-standing disagreement between Glaser and Strauss about the methodology. This resulted in them producing separate publications on grounded theory and how it should be carried out, and their students also contributed to the evolution of grounded theory (Walker & Myrick, 2006). Researchers have a number of grounded theory methods to choose from (e.g. Glaser and Strauss, 1967; Strauss and Corbin, 1998; Charmaz, 2008); however, all types of grounded theory provide useful guidelines for the collection and analysis of data (Charmaz, 2009).

2.1.4 Constructivist Grounded Theory

It was decided to use Charmaz's (2008) constructivist grounded theory approach for the present study. Charmaz (2009) describes this approach as a modern version of Glaser and Strauss (1967) and Glaser (1978). However, Charmaz (2008) does not assume that theories emerge from the data distinct from the researcher and instead proposes that they are a construction of reality between the participant and researcher;

"...we are part of the world we study and the data we collect. We construct our grounded theories through our past and present involvements and interactions with people, perspectives and research practices" (p. 10).

In constructivist grounded theory, data analysis is perceived as an interpretation and not a single viewpoint on the research area and the importance of reflexivity is highlighted (Charmaz, 2009). The acknowledgement of the researcher's impact on the entire research process and the reflexive nature of Charmaz's approach made it an appropriate choice for use in the present study. Working in the profession of clinical psychology, the researcher values the importance of being mindful of her impact on clients within the therapeutic relationship, which transfers well to the constructivist grounded theory approach:

"The grounded theorist's analysis tells a story about people, social processes and situations. The researcher composes the story; it does not simply unfold before the eyes of an objective viewer. This story reflects the viewer as well as the viewed" (Charmaz, 2000, p.522)

Furthermore, Charmaz (see Charmaz, 1999) has carried out numerous studies on the experiences of people with chronic illnesses. Although in this study the participants have not experienced a chronic illness they have faced a health challenge in terms of

being at risk of a chronic, degenerative disease and are often living with someone with HD. Other researchers have also used constructivist grounded theory to explore how people cope with health problems such as heart disease (e.g. Falk, *et al.* 2007) and how relatives manage the grief associated with losing a spouse to cancer (Holtslander & Duggleby, 2009).

2.2 Ethical Considerations

The main ethical issues in the present study related to protecting the identities of participants because of the secrecy and stigma that is often associated with HD. It was important to be mindful that participants may have relatives who are unaware of the disease risk within their family and to respect this throughout the duration of the study.

2.2.1 Anonymity

In order to ensure anonymity, code numbers were used instead of participants' names on interview transcriptions. Documentation linking participants' names with their designated code number was saved on a password protected NHS computer. Participants were informed verbally and in writing that direct quotes would be used in the written report of the research to illustrate themes; however, any personally identifiable information (names of people, places, dates etc) would be altered or removed.

2.2.2 Confidentiality

A number of steps were taken to ensure confidentiality. The research interviews were transcribed by the researcher and the recordings were deleted once transcriptions were

complete. The anonymised transcripts were stored in a locked filing cabinet in the researcher's office. The consent forms were kept in the researcher's office in a separate locked filing cabinet to the transcripts.

2.2.3 Informed Consent

The participant information sheet (see Appendix 1) highlights that participation is voluntary, participants are free to withdraw from the study at any time without having to give a reason, that there is no impact on care regardless of whether an individual decides to participate or not, potential risks of participation and that there are no direct benefits associated with taking part in the study. At the beginning of each research interview participants were reminded of the aim of the study, what participation would involve and their rights were explained. Participants were also given the chance to ask questions relating to the research and their involvement in the study. Once participants indicated they were willing to take part in the research they were asked to sign the consent form (see Appendix 2). Participants were also requested to provide consent for their interview to be audio-recorded.

2.2.4 Support for Participants

Given the sensitive nature of the research it was possible that some participants could become upset during the interview perhaps due to talking about issues relating to other family members with HD. If this happened the interview would stop and the person would be asked if they would like a break and whether they would like to continue. If there were concerns about a participant's emotional state either during or after a research interview then support would be offered. If required, support or advice would

be provided by healthcare professionals with expertise in HD and genetic counselling. In addition, participants were informed in advance of the research interview that if the researcher had serious cause for concern for their safety or well-being then it would be in their best interest for the researcher to inform an appropriate healthcare professional. If there were severe clinical concerns that required immediate intervention e.g. suicide risk then external services would be contacted e.g. Liaison Psychiatry. A participant would be made aware of any contact made with a third party regarding their situation.

2.2.5 Ethical Approval

An application for ethical approval was submitted to the Research Ethics Committee of the Doctorate in Clinical Psychology at the University of Edinburgh and the study was granted permission to proceed (see Appendix 3). Ethical approval was then sought from the North of Scotland Research Ethics Service (ref: 09/S0801/39). Following a response from the committee, a couple of minor changes were made to the participant information sheet and approval was obtained (see Appendix 4). In addition, the local NHS Research and Development Department provided management approval for the study (see Appendix 5).

2.3 Procedure

Following ethical approval it was possible to start participant recruitment. The inclusion criteria for the study were people aged 23 years or over who received a favourable result from predictive genetic testing for HD at least five years ago (minimum age for participating in genetic testing is 18). The research interview required participants to speak at length about their experiences therefore individuals

with a poor understanding of English were excluded from the study. If potential participants expressed that they did not want their interview to be audio-recorded then they were excluded from the research. Recordings were necessary for the purposes of detailed data analysis.

2.3.1 Sampling

The population of interest in the present study could be described as ‘hard to find’ in the sense that HD is a relatively rare disease, the numbers of people who actually go for predictive testing are quite low, and the numbers receiving favourable results are fewer still. A further limit on sampling was that the study required participants who had their test result at least five years ago. In a grounded theory study, researchers start with selective sampling which means choosing which locations and populations to sample from before data collection begins, and then the researcher progresses to theoretical sampling as codes and categories begin to develop (Draucker *et al.* 2007).

Charmaz (2008a) defines theoretical sampling as “*seeking and collecting pertinent data to elaborate and refine categories in your emerging theory*” (p. 96). Although theoretical sampling is the main sampling strategy used in grounded theory, the researcher was limited to one setting for recruiting participants and due to the small population there was no opportunity to specify particular characteristics of participants in line with developing categories (e.g. wanting to select a participant who already had children when they found out they were at risk of HD). However, theoretical sampling was still possible, because with each new participant the researcher explored developing categories and was able to identify further properties and dimensions of these

categories. Charmaz (2008a) explains that it is incidents and events that are the focus of sampling and not participants. Furthermore, Strauss and Corbin (1998) state that differences in data often develop due to the natural variations than occur in incidents and events. Although in the present study the researcher did not have control over selection of participants, the sample comprised a variety of individuals with different backgrounds, personal circumstances and experiences of HD and predictive testing. Due to the variation across the participants' experiences, there was the opportunity to explore developing categories within different contexts and for comparisons to take place between incidents and categories.

2.3.2 Participant Recruitment

The participants in the study were recruited through a Consultant Clinical Geneticist with a specialist interest in HD. During her long career she has delivered over 200 predictive test results to people at risk of HD, of which approximately half were favourable. She identified potential participants by both her own knowledge of past patients and by also reviewing her departmental patient records. She only approached people who were at least 5 years post-test result and who met the inclusion criteria. Invitation letters (see appendix 6), participant information sheets, contact details forms and stamped addressed envelopes were sent by the Consultant to potential participants. Any individuals who were not considered appropriate for participation in the study were not approached.

Potential participants who were interested in taking part in the study were asked to return a contact form to the researcher in an enclosed stamped-addressed envelope.

The contact form requested the potential participant's name, address and telephone number and whether they would like to be contacted by the researcher by telephone or letter. When this was received contact was made with the potential participant by their preferred method. For those who wanted to be contacted by letter, the letter thanked them for their interest and requested them to phone the researcher if they wanted to take part in the study and to arrange a suitable date and time for the interview. For those who wanted to be contacted by telephone, they received a phone call to arrange a date and time for the interview. The researcher spoke to all participants on the telephone prior to the research interview. This provided an opportunity for the participants to ask questions and to get to know the researcher. Efforts were made during the conversation to put the participants at ease and build rapport ahead of meeting them in person. At the start of the interview participants were asked to complete the consent form. The recruitment period lasted for approximately 6 months interwoven with data collection and analysis. Studies using grounded theory can typically have sample sizes ranging from 10-60 participants (Starks & Trinidad, 2007).

2.3.3 Characteristics of the Participants

The sample comprised ten people who had received a favourable result from predictive testing for Huntington's disease. Six women and four men participated in the study and their ages ranged from 42 to 62 years old (mean age 53.4 years). Six participants were married, three were divorced and one participant was single. The length of time participants were aware of their risk of HD ranged from 7 to 50 years (mean 24.5 years) and the age at time of receiving the predictive test ranged from 26 to 54 years (mean

42.2 years). The length of time since receiving a favourable result ranged from 5 to 20 years (mean 11.2 years).

Table 1: Participant demographics

	Age	Gender	Marital Status	Length of Time Known Risk of HD	Age at time of Predictive Genetic Testing	Length of Time Since Favourable Result
PARTICIPANTS	59	Male	Married	50 years	39	20 years
	44	Male	Single	15 years	34	10 years
	58	Male	Married	11 years	53	5 years
	62	Female	Married	40 years	44	18 years
	42	Female	Divorced	15 years	28	14 years
	52	Male	Married	10 years	47	5 years
	53	Female	Married	37 years	45	8 years
	58	Female	Married	30 years	52	6 years
	46	Female	Divorced	30 years	26	20 years
	60	Female	Divorced	7 years	54	6 years

2.3.4 Data Generation

In a grounded theory study, data can be generated from many different sources such as observations, individual and group interviews, documents, television programs, diaries and other first-hand accounts (Schreiber, 2001). The research question or aim can influence the methods of data collection a researcher selects (Charmaz, 2008). Given the sensitive nature of the research topic it was decided that in-depth interviews would be the most appropriate and informative method of data collection. Prior to beginning data collection, discussions were held with the Consultant Clinical Geneticist involved in the research to ask her advice on interviewing people who have received favourable predictive test results for HD. Having very little experience in the area, the researcher wanted to have knowledge of the genetic testing process and also to be aware of potentially difficult issues that may arise for the participants during the interviews. The

Consultant's extensive clinical experience in this area was invaluable in preparing the researcher for the interviews. It was important for the researcher to keep this information in mind yet allow the participants to speak about what was important and meaningful to them.

2.3.4.1 Semi-structured Interviews

Participants took part in a research interview lasting approximately one hour. The interviews occurred in the researcher's office in an NHS setting. The researcher was mindful that participants may feel nervous and time was spent at the start of the meeting chatting about general conversation topics, gaining rapport and explaining what the interview would involve. At the beginning of the interview, participants completed the consent form and also provided demographic information. It was anticipated that this would help the participants relax.

An interview guide was created (see appendix 7) following the advice and guidelines of Charmaz (2008a). She provides a list of sample grounded theory interview questions relating to a life change which is relevant to the present study. Some of the sample questions were adapted for use within the present study. However, the questions were not referred to routinely with each participant and instead a flexible approach was adopted. The guide was useful in terms of thinking about how to phrase questions and helped particularly when participants were less verbose or when the researcher was anxious and struggled to think of an open-ended question during the initial interviews. Charmaz (2008a) states that an interview guide can help to increase researchers' confidence and allow them to pay attention to the participant's story. The questions

were referred to more in the early interviews and were used less as data collection progressed, as developing categories were being explored with participants and the researcher's confidence increased. The researcher started each interview with the question: "I am interested in hearing your experiences of receiving a favourable result from predictive testing for HD. Please start wherever is easiest for you?" All of the participants started the interview at the beginning of their HD story in terms of talking about when they first discovered they were at risk of the disease, deciding about going for predictive testing, going through testing, receiving the result and then life after the favourable result. Nunokoosing (2005) explains that research participants select components of their lives that they have the greatest interest in conveying, and in his opinion all of the experiences a participant decides to divulge are on a par in terms of the importance of their contribution to the researcher's understanding of their world.

In relation to the present study it was noticeable after the first few interviews that participants were keen to talk about finding out about their risk of HD and the subsequent genetic testing process. Although the initial research aim was interested in life after receiving the favourable result it emerged that it was important and meaningful for participants to tell their HD story from the beginning. It was discovered that an understanding of life after the result was not possible without an appreciation of the journey and life context up to that point.

At the end of each research interview the researcher made attempts to finish on a positive note. Participants were asked if they had anything else they wished to include in the interview and they were thanked for taking part in the research project. Although

the participants were reminded that support was available for them, no one requested it and general feedback from the participants indicated that the interview had been a positive experience.

2.3.4.2 Demographic Information

Demographic information was requested from participants for the purposes of describing the sample and providing a context for the study. Participants were asked to provide details of their gender, age, marital status, how long they had known they were at risk of HD, and how long since they had received their favourable result.

2.3.5 Data Analysis

The interviews were transcribed verbatim by the researcher. Each interview document contained a wider right-hand margin to leave space for codes. The lines of the transcribed interview were numbered to make it easier to keep track of codes and to find quotes to back up the codes. Although aware of the advantages of using computer software packages for qualitative data analysis the researcher decided against this option in order to allow for deeper engagement with the data. Charmaz (2000) reported her doubts about using such programs and stated that they are more appropriate for objectivist grounded theory than constructivist. The findings were analysed manually following the coding procedures of Charmaz (2008a). Coding is described as;

“the pivotal link between collecting data and developing an emergent theory to explain these data. Through coding you define what is happening in the data and begin to grapple with what it means” (Charmaz, 2008a, p.46).

Initial coding was carried out with the first three interviews and this involved a mixture of line-by-line coding and incident-by-incident coding. In keeping with Charmaz’s (2008b) recommendations the codes were short, specific and active in order to help highlight processes in the data. An example of initial coding from Interview 03 is presented in Figure 1:

Interview Transcript

Initial Coding

03: <u>I came back to see Dr X and I mean...the way she does it...I</u>	changing mind
<u>felt is that it sort of took me on some sort of a journey</u>	starting a journey
<u>almost....that I wondered if I wanted to do this.</u> By the time I got	feeling unsure
to the point of blood tests, actually some had been taken, you	
know <u>I had moved from where I was to a place where I really</u>	feeling ready
<u>was sure that I wanted to know the result</u> and erm....I mean the	making the decision
issue for me was that if I didn't have this disease <u>I didn't want to</u>	doing it for self
<u>spend the rest of my life worrying about it and I didn't want my</u>	doing it for my children
<u>kids to have to you know spend their lives worrying about it.</u>	

Figure 1: Example of Initial Coding

Incidents were compared both within and across the three interviews to identify similarities and differences between codes and concepts. Making comparisons and asking questions of the data helped to identify properties and dimensions of emerging concepts (Charmaz, 2008b). After coding the first three interviews, ideas about the data were developing. For example, the participants appeared to want to tell their story from the beginning of their HD experience which was when they found out they were at risk of the disease. In relation to this, similar codes were grouped into higher order

concepts such as “finding out about the risk”, “living with the uncertainty” and “making the decision”. This appeared to set the context for what happened after the participants received their favourable result and there was a sense from the data that participants were “trying to distance themselves from HD” but they varied in how successful they were in doing this. These emerging concepts and categories were explored in subsequent interviews. Focused coding was then engaged in and this involved using the most common and important codes from the initial interviews to sort through the data (Charmaz, 2008b). Incident-by-incident coding was also used in these interviews which created further codes.

As analysis progressed, theoretical coding and continued memo writing helped in starting to explore relationships between categories. Two main categories were identified which were “facing a journey into the unknown” and “trying to distance self from HD”. These central processes and their subcategories explain the difficulties people at-risk of HD face from the moment they find out about the risk through to receiving their favourable result and then trying to move forward with their lives. Only one participant was able to report that he has been able to forget about HD since receiving his favourable result. This negative case helped to add explanatory power to the substantive theory. A detailed presentation of the results can be found in the next chapter.

2.3.5.1 Memos

Memos are a very important part of data analysis in grounded theory. Writing memos helps to provide direction during coding and also for breaking down categories into

properties and dimensions (Charmaz, 2008b). Memo writing encourages researchers to begin the analysis of codes early in the process with ideas and codes becoming more abstract through continuing to write memos as data collection/analysis progresses (Charmaz, 2008a). Memos written early in the analysis process are used to explore and expand codes which then helps to direct subsequent data collection, whereas more advanced memos written later in the analysis process are used to describe how categories develop and change and to make comparisons. Memos also provide the opportunity to compare data with data, incidents and categories both within and across interviews (Charmaz, 2008a). In the present study memos were written throughout the research process and helped to keep track of developing ideas, describe categories through identifying their properties and dimensions and explore their relationship to each other. Memos were also helpful for generating questions about codes and categories to explore in future interviews. Figure 2 is an example of a memo written early in the process:

Memo – Finding out about the risk of HD

The point at which people realise they are at risk of HD is a significant life moment/start of a journey into the unknown which introduces uncertainty about their future. Some people find out about risk when another family member has become unwell and been diagnosed with HD with no obvious previous family history of HD – other people know from a young age as they have grown up with a parent who has HD. How do these perspectives differ in terms of coping with the risk of HD? Life circumstances at time of finding out about the risk – some people were married with children and others were single – different implications for people. Feelings at time of finding out include fear, shock, powerless - explore this in other interviews

Figure 2: Example of Memo

2.3.5.2. Reflective Journal

During data collection and analysis, reflective notes were written as a way of considering the impact which the research was having on the researcher and vice versa. Thoughts and feelings about the interviews were also recorded. Figure 3 is an example taken from the reflective journal.

I have carried out a few interviews now and am finding that I am more able to relax. I am worrying less about asking the right questions and am not looking at the interview guide as much. I feel that I can focus more easily on what the participants are saying and am asking more open questions instead of closed questions with yes/no answers.

Figure 3: Excerpt from Reflective Journal

2.3.5.3 Saturation

Saturation is defined as the stage: “*when gathering fresh data no longer sparks new theoretical insights, nor reveals new properties of your core theoretical categories*” (Charmaz, 2008a, p.113). It was decided to stop collecting data after 10 interviews as it was felt that the data was approaching saturation. It is debatable as to whether saturation can ever be truly achieved as it is always a possibility that categories can be adapted (Willig, 2008). Nevertheless in the present study the final interview did not reveal any new categories and it was felt that the categories that had emerged from the data provided a meaningful account of the main problem for the participants.

2.4 Validity

As qualitative research is relatively new to psychology, it is important for qualitative researchers to demonstrate rigour in their research and produce findings that are on a par with quantitative research in terms of their usefulness (Yardley, 2008). Yardley (2000) developed a set of principles which qualitative researchers can use to ensure quality in their research; sensitivity to context, commitment and rigour, coherence and transparency and impact and importance. Yardley (2000) states that the principles should not be perceived as rules that have to be obeyed and they can be interpreted flexibly which is in keeping with qualitative research methodologies. Despite this it is still important that researchers provide justification for any deviation from standards for good research practice (Yardley 2008). The validity criteria of Yardley (2000; 2008) were referred to in the present study as follows:

2.4.1 Sensitivity to Context

Sensitivity to context involves the researcher being aware of the importance of the context of theory, the socio-cultural context of the research setting, and the context of the relationship between the researcher and participants (Yardley, 2000). In relation to the present study, sensitivity to the context of theory was demonstrated by carrying out a review of the literature on the psychological consequences of predictive genetic testing for HD. In addition, qualitative research studies in similar research areas were examined, for example predictive genetic testing for hereditary breast cancer. The review of the literature highlighted the need for further qualitative research exploring the experiences of people who receive favourable results from predictive genetic testing for HD. Detailed discussions were also held with a Consultant Clinical Geneticist who

has extensive clinical and research knowledge and experience in the field of HD and predictive genetic testing. This enabled the researcher to become familiar and sensitive to the genetics terminology, the nature of HD as a disease, the process of predictive genetic testing for HD, complex ethical issues facing families at risk of HD and previous research in the area.

Sensitivity to the socio-cultural context of the setting was considered through deciding not to hold the research interviews in the Clinical Genetics department. Although this was a familiar setting to the participants, most or all of them had not been there since receiving their favourable test result at least five years ago. Revisiting this location could have triggered memories of a distressing time and also potentially seeing patients with HD could be upsetting given the presence of the disease in their family. Participants had the option of taking part in the research interview at home or in the researcher's office which is in an NHS rehabilitation hospital setting. The hospital is at a different site to where the participants would have attended for predictive genetic testing. All of the participants chose to be interviewed in the researcher's office which was made as relaxing and inviting as possible.

The relationship between the researcher and the participants was considered throughout the study from the design through to data analysis. Prior to beginning the study, the researcher had very little knowledge and experience of HD or predictive genetic testing. As a result the interviews were flexible in nature in order to allow the participants to tell their stories in their own words and discuss what they thought was important and meaningful to them. Although it is difficult to deny the power imbalance between the

researcher and the participants, (Yardley, 2000) it was hoped that by not having a structured set of questions prepared and instead adopting a flexible and open questioning approach, this would communicate a genuine interest to the participants and make them feel at ease. The participants were aware that the researcher had professional links with the Consultant Clinical Geneticist who had delivered their test result and it is possible this influenced their responses during the interview. Most participants were, however, very open and honest in providing their views about the genetic testing process. The researcher was also very mindful of the sensitive nature of the research and the fact that participants could have relatives in the advanced stages of HD or could have lost family members to the disease.

2.4.2 Commitment and Rigour

Yardley (2000) defines commitment as:

“prolonged engagement with the topic (not necessarily just as a researcher, but also in the capacity of sufferer, carer etc) the development of competence and skill in the methods used, and immersion in the relevant data (whether theoretical or empirical)” (p.221).

The researcher demonstrated commitment through conducting a detailed literature review of the topic and also carrying out background reading on HD and how it affects families. In addition, the researcher has previous experience of using qualitative research methods and has attended workshops and training on grounded theory. The research interviews were personally transcribed by the researcher which enabled her to immerse herself in the data and become familiar with the participants and their experiences.

In terms of rigour, which Yardley (2000) defines as; “*the resulting completeness of data collection and analysis*” (p.221), this was achieved by triangulation of data whereby data was collected from participant interviews, detailed discussions took place with a Consultant Clinical Geneticist and the literature was consulted in order to compare the study findings with previous theories or models in the area. Although the participants in the study could be described as a hard to reach population due to the relative scarcity of people who go for predictive genetic testing for HD, variation in the sample was still achieved. For example there were variations in age, gender, time since first aware of risk of HD, length of time since receiving a favourable result, marital status, and the effect of HD on the family. The analysis procedures described earlier enabled the researcher to engage in-depth with the data and develop a theory that was grounded in the experiences of the participants.

2.4.3 Coherence and Transparency

In order for a study to have coherence then it must be clear how the research aim fits with the research method and furthermore it is important to have adequate knowledge about qualitative research methods in order to select the most appropriate method for the study (Yardley, 2000). It has been justified earlier in this chapter why qualitative research and constructivist grounded theory in particular were chosen to meet the aim of this study. Also the researcher has background experience and knowledge of qualitative research and this helped when considering which approach to use in the present study. It had been planned to invite two participants from the original sample to give their views on the substantive theory in order to find out if it was meaningful to them and reflected their experiences. However, due to time restrictions unfortunately this was not

possible. Copies of the findings will however be sent to participants who requested this information and they will be invited to provide feedback. The researcher also regularly returned to the raw data to check the developing theory was grounded in the experiences of the participants. The coherence of the analysis was interrogated through discussions with the Consultant Clinical Geneticist involved in the project and also during research supervision.

Transparency is about the researcher being open and honest in relation to how the study was carried out so that other people can have a clear idea of the methods involved and why the researcher chose them (Yardley, 2000). Transparency can be achieved by providing an audit trail and also through reflexivity (Yardley, 2000). An audit trail was kept throughout the data analysis process in the form of coded transcripts, reflective notes about interviews and the analysis process, memos detailing the development of categories and their properties and dimensions, and diagrams of relationships between categories. In terms of reflexivity, the researcher was mindful of how working as a trainee clinical psychologist could influence how she interpreted the data. She was open in her approach to the data and did not use preconceived psychological concepts to code the data.

2.4.4 Impact and Importance

The idea to carry out the present study came from discussions with a Consultant Clinical Geneticist about predictive genetic testing for HD. She raised the issue that some people who receive favourable results from predictive testing struggle to adapt to the news that they are no longer at risk of the disease. The consultant was of the

opinion that there had been little research activity in this area and that it was worthwhile exploring it further. The results of this study would be of interest to both health professionals working in the areas of genetic counselling and clinical health psychology in terms of explaining the psychological consequences of receiving a favourable result from predictive testing for HD and informing psychological interventions.

2.5 Chapter Summary

In summary, this chapter has justified choosing grounded theory for the present study and has described the main features of the approach, before considering the ethical issues relating to the research. The procedure was outlined including sampling, participant recruitment, data generation and analysis, and the techniques use to ensure validity of the findings were discussed. The next chapter presents the findings of the study.

Chapter 3

Findings

3.0 Introduction

The initial aim of the study was to explore the experience of receiving a favourable result from predictive testing for HD and how this has impacted on relationships in the long-term. Although the research was interested in the time period after a person received the favourable result, it became clear early on in data collection/analysis that participants wanted to tell their story from the moment when they found out they were at risk of the disease. This highlighted how important and meaningful these experiences were in their overall journey through predictive testing and beyond, and helped to provide the context for understanding their experiences after receiving the favourable result. Therefore instead of ignoring this discovery it was decided to embrace the participants' desire to speak about events and experiences leading up to their favourable result in addition to life afterwards.

3.1 Overview of the Findings

A substantive temporal model (see Figure 3) has been created which has two major categories; Firstly, *facing a journey into the unknown* describes the process participants went through when they found out they were at risk of HD. Within this process, after *finding out about the risk* participants appeared to vary in their ability to *manage the uncertainty* associated with living at risk of HD. *Making the decision* to be tested was

the first step for the participants in moving towards removing this uncertainty and ending their journey into the unknown.

The second major category *trying to distance self from HD* explains the time period after participants found out they did not have the disease. After receiving the result and *facing the truth*, participants experienced a variety of emotions. For some individuals, positive feelings were often short-lived due to the experience of *breaking the news* to family members who either had HD, or were still living at risk of the disease. *Moving forward* after receiving the favourable result was not straightforward for most of the participants and their success in getting on with life depended on various factors. Participants who had no close family members with HD or had emotional distance or geographical distance from family members with HD or at risk of the disease, were more able to ignore HD and live a normal life again. Participants who had siblings or a parent with HD found it more difficult to leave HD behind. There was also the problem of *dividing loyalties* for those participants who had children and were torn between getting on with normal life with their immediate family and also having a sense of duty to their extended family with HD.

The category *having a sense of duty* explains the process which prevented some participants from distancing themselves from HD. It describes the experiences of those participants, who because of their sense of duty to their family members, they are still living with the disease in their lives. Caring for siblings or a parent in the advanced stages of HD, fighting for the rights of family members with HD and giving something back through charity work are all examples of having a sense of duty to family with

HD. Quotes from participant interviews are used throughout this chapter to illustrate examples of categories and subcategories and to ground the data in the participants' experiences.

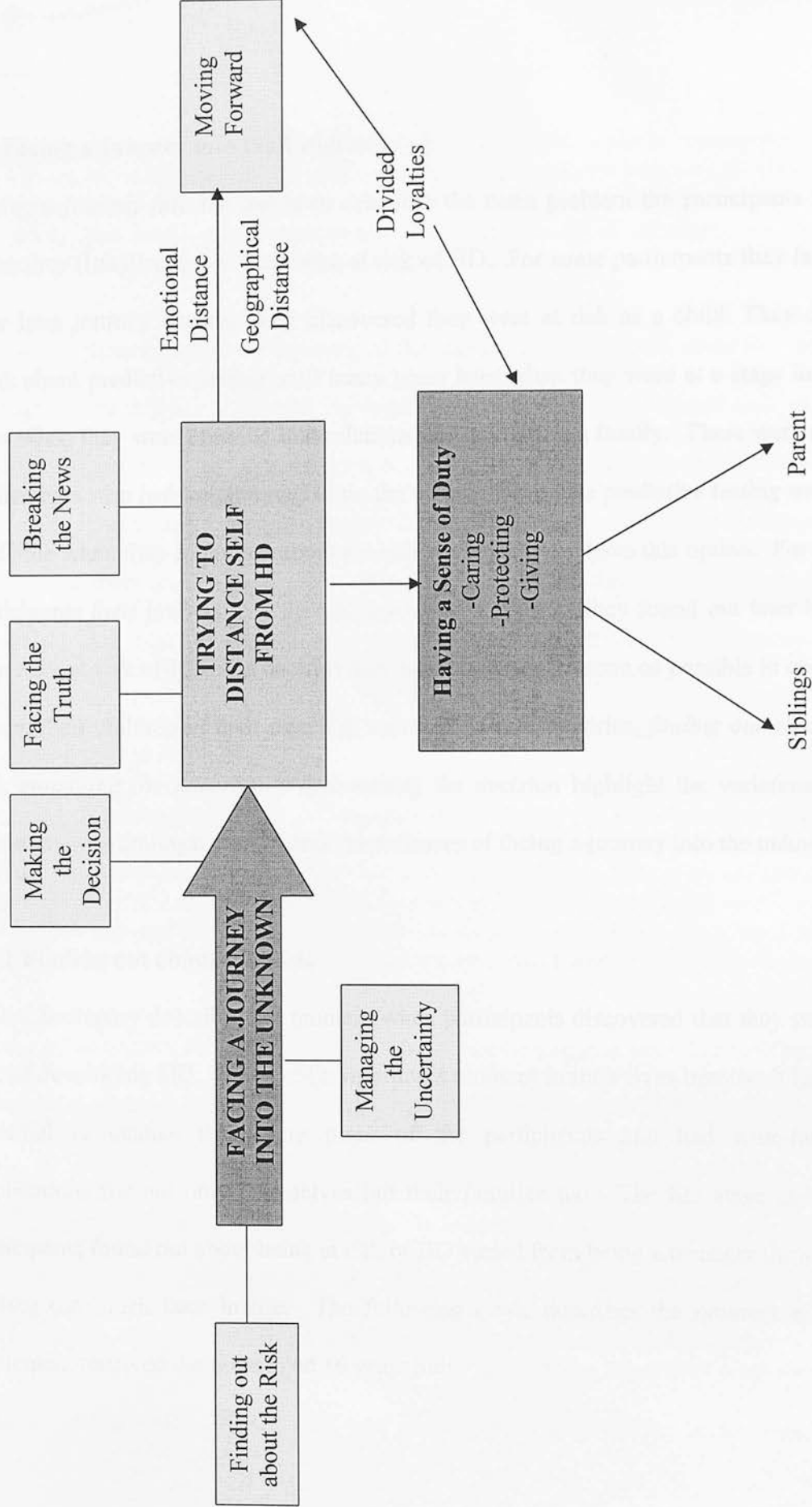


Figure 4: Substantive Model

3.2 Facing a Journey into the Unknown

Facing a journey into the unknown describes the main problem the participants faced when they first discovered they were at risk of HD. For some participants they faced a very long journey because they discovered they were at risk as a child. They didn't think about predictive testing until many years later when they were at a stage in their lives when they were entering into relationships or starting a family. There were a few participants who had long journeys into the unknown because predictive testing was not available when they found out about the risk so they did not have this option. For other participants their journey into the unknown was shorter as they found out later in life they were at risk of HD, and decided they wanted testing as soon as possible in order to inform their children of their own risk status. The subcategories, *finding out about the risk*, *managing the uncertainty* and *making the decision* highlight the variations and commonalities amongst participants' experiences of facing a journey into the unknown.

3.2.1 Finding out about the Risk

This subcategory describes the moment when participants discovered that they were at risk of developing HD. It marked a significant moment in their lives because it had the potential to change the future plans of the participants and had wide-ranging implications for not only themselves but their families too. The life stage at which participants found out about being at risk of HD varied from being a teenager through to finding out much later in life. The following quote describes the moment when a participant received the news aged 16 years old:

03: *Erm well it started I suppose, well I might as well start at the beginning which was when I was 16 which was when my father and....it was my father who was at risk himself at the time....told us at that time that his mother had got Huntington's and erm....this was a big...you know, a big event in our family life, he sat us all down and he was pacing up and down the room and really distressed you know that he had to tell us.*

The above quote captures the intensity of the moment of finding out about the risk and highlights how difficult it was for the father to pass on the news that not only he is at risk of the disease but his children are too. The following participant was aware of HD from a very young age due to her mother having the disease and being symptomatic for much of the participant's childhood. There was a sense from the participants who found out about their risk at a young age that HD wasn't something to be immediately concerned about:

09: *Erm I was brought up with HD because my mum had HD erm and I was told from a young age by my father that I wasn't allowed to have children because of carrying HD and as I has a brother as well he was also told the same. A lot of my aunties on my mum's side got HD as well. My mum died when I was just coming up to 13, of HD. And then it was just like a part of my life but came more into my life when I started maturing and then I went into a steady relationship when I was in my early twenties.*

On the other hand some participants found out about their risk later in life when they already had children which meant that their children and grandchildren were also at risk:

08: *...but anyway basically from there at that stage erm I was married, I've got 3 children erm.....my oldest lad is now 24 but my daughter at the time, she was about 18 or thereabouts. Unfortunately she was in the situation where she had fallen pregnant...*

The participants appeared to experience a variety of emotions when they were told they

were at risk of HD. Feelings of fear and anxiety were typical of the emotional

responses of some participants and the fear appeared to relate to both the self and others:

01: Well away back when we first found out about it.....it was....what's the word I'm looking for erm...scary....I had one son and was pregnant with the other one and I knew my uncle was going back and forth to hospital for different tests until we finally found out what it was...

Participants who found out they were at risk of HD shortly after family members were diagnosed with HD seemed to find the news more anxiety-provoking. The family members had been displaying a variety of symptoms including behavioural issues for some time before the diagnosis. Finding out about the risk of HD seemed to be more difficult for those who had family members in the advanced stages of the disease and they had observed the impact which it has on the family. It is possible that the anxiety related to the fact that the participants could potentially end up the same way as their family members and this created feelings of fear.

05: ...but until you actually experience it you can't possibly imagine how it is going to impact on your family. It wasn't until mum started showing symptoms that you realise how it does impact not just on how mum is but it is the impact on my relationship with my sister and my brother, my relationship to my husband at the time, everything really.....

In contrast, there were some participants who appeared not to worry so much about being at risk. A few participants had relatively positive past experiences of HD in their families so therefore had less anxiety about being at risk of the disease:

02: My father was diagnosed with it and although he didn't die of it, it was bowel cancer that he died of...eh...and it was, it was Dr X she eh explained that I could have it,

and my sisters and brothers they could have it as well...eh...well there was nothing I could do about it really...I mean if there was medication or something towards it...until then you just have to live with it really...because he wasn't actually too bad, my father yeah... So we were just fortunate.....that eh.....if...well he, he died, it was fortunate that we could see that, that it was going to be similar to that, well we were hoping for that anyway....

This participant appeared hopeful that if he did have the HD gene then the disease would affect him in a mild way as it did with his father. Similarly, the following participant suggested that she wasn't very emotional when she found out she was at risk and did not see HD as something to be immediately worried about. Again her family experience of HD hadn't been too distressing, the onset had been late in life and she had not witnessed the effects of HD at that stage in her life:

03: I was remarkably matter of fact about it. I don't think it is terribly real to you....sorry to me. To my sister it felt much more real but to me it wasn't especially real. This was something that might happen at some point in the distant future because the other thing about my family is that my grandmother didn't get it until very late so, so, and I had never seen my grandmother after she got it. It, it, it, wasn't terribly real to me.

An interesting feature of finding out about the risk was the means by which participants discovered the news. There was considerable variation between the participants, for example there were some individuals who knew from a young age about their risk of HD because they were living with an affected parent. On the other hand there were a few participants who had a parent die relatively early in life before the onset of HD and the family therefore did not know the gene was in the family. It was only when later in life when siblings of the participants started showing symptoms which then led to the diagnosis of HD, and the realisation that they were at risk.

08: *It was my brother who actually has Huntington's, and erm initially they actually thought he had MS erm and they weren't very sure for a long time. He went through quite a lot of tests and then there was a chap from X Hospital who worked for the genetics department and he got involved. He actually came to my brother's house to interview him and what he found was that my brother showed all the classic signs of Huntington's, Anyway my mother was tested for HD and she.....didn't carry the faulty gene either. No, unfortunately my father, he died in 1952, and I was about 18 months old, he was 35 years of age so we believe that HD has actually come through that side of the family.....*

3.2.2 Managing the Uncertainty

After finding out about the risk of HD participants had to try to manage the uncertainty related to facing the unknown. The length of time which the participants lived at risk before going for predictive testing varied from less than a year to over thirty years. It is possible that those who lived with the risk for the longest period were more able to tolerate the unknown. This subcategory illustrates how the participants tried to manage this uncertainty ranging from those who tried to ignore HD and get on with life to those who couldn't switch off from thoughts of HD and tended to over monitor their behaviour for symptoms of HD. A few participants tended to fear the worst and would search for signs of HD in themselves:

01: *Oh aye, for years – if you had anything, if you dropped, just the slightest thing you thought – this is the start of it...*

10: *... somehow or other when a door is open I will always seem to manage to walk into the side of the door – bang!*

These participants seemed hypersensitive to any sign that they may have HD and were living life as if they were waiting for HD to start. In addition to looking for physical signs that she may have the disease, participant 01 also tried to search desperately for clues that she may not have the positive gene by comparing herself to her father who didn't have HD:

01: ...and all I kept saying to myself was I'm more like my father, I've got more of his genes and I am more like my father but eh you've genes from the pair of them, ye ken that...

The following participant appeared to struggle to switch off from thoughts of HD and explained that since finding out about the risk there was no turning back:

05: You know she (mother with HD) didn't know about it (HD) whereas we were aware of the risk and she was completely oblivious to it, you know she wasn't aware there were any risks or any potential impact on the family whereas once you have got that knowledge it is very difficult to ignore it isn't it.....

In contrast, other participants were more able to get on with their lives and to an extent ignore the threat of HD. It appeared as though they had accepted their risk status:

04: I can't say that I've worried myself that I might have it....you know there is always the possibility but you've just got to get on with life....I wasn't like dropping a cup and thinking oh my goodness. I'm not that kind of person anyway but I imagine some people would...

09: I never actually really thought about it. It didn't, I think it prayed on my brother's head a lot more than it did mine. I just accepted that well one day if I wake up not well, I wake up not well and if I don't, I don't. I could die from something else before then, I could get hit by a bus.....my aunts and uncles made it – it was a big massive thing, oh my god you are going to wake up one day when you are thirty and you are not going to manage to get out of your bed....and I thought well today I'm fine and today I'm doing this and that's just the way I coped with it.....

Living one day at a time appeared to help participant 09 manage the uncertainty of being at risk of HD and she appeared to try to put her risk into perspective. Similarly, the following participant also tried to get on with life and gain some perspective on her situation by listing all the other life worries that people face. She appeared able to not let HD completely dominate her mind and her future plans:

03: *Yeah.....so quite frankly I was extraordinarily fortunate because I.....I don't actually remember telling my boyfriend, who is now my husband, erm about it but I mean he just took it on board that this was a risk like walking under a bus is a risk you know he accepted it and then as time passed we decided that we would have children...erm that primarily erm.....you can't spend your life worrying about something that may not happen. You have to get on and live your life.....and I always felt that there were too many other things to worry about in life actually, you know worrying about earning enough, worrying about what you are going to do with your life in terms of jobs and all these things, to be too focused on Huntington's.*

Having a supportive partner clearly helped the above participant and they were both able to accept the risk and move on with their major life plans such as getting married and having children without letting HD dictate their lives. Of the participants who didn't already have children when they found out they were at risk and had to consider family planning issues thereafter, half went on to have children without being tested and the other half waited until they received their favourable result. One participant had four children whilst still at risk of HD and it is only years later when reflecting back that she realised what could have happened. It is possible the participant and her husband had children because it felt like the right thing to do at the time and they didn't want to let something that may happen in the future spoil their plans:

03: *...my mum deeply disapproved of this. I think she felt, she felt that having two children was maybe you know okay, but to actually have four children, I mean lots of people disapprove of having four children in any circumstances, let alone when you've got a risk of Huntington's disease. I mean it was a pretty stupid thing to do actually when I think about it but anyway that's the way we did it*

Both participant 03 and participant 09 had to face the disapproval of family members when they had children whilst still at risk of HD. It was apparent that their parents did not want them to risk potentially passing on the HD gene to another generation:

09: *When I got married I was adamant that I wanted children and I fell pregnant within a couple of months of being married. This didn't go down too well with my father at all and it was like – what are you doing? I was like, well it is my life I will deal with it the way I want to do it. My dad couldn't understand after seeing my mum with it and dying so young with it how I'd want to put myself or my daughter through something like that. My argument was that there was still a 50-50 chance that I don't have it and my daughter won't have it and my dad says well that is still 50-50. And I says well you took the chance so.....I'd always wanted to have children from knee high*

Having a strong desire to have children prevented her from going for testing initially as she knew she wouldn't have gone ahead with having children if she had HD. It was only after she became a mother for the first time that she then felt ready to go for testing:

09: *I'm glad that I don't have it because if I had, had it I would've got sterilised and I wouldn't have had no more kids – I would have just had the one. I think it was just more the fact that I wanted to be a mum, then I was a mum and then I thought if I had it I would've went ahead and got sterilised, much that it would have pained me to do so but I could have said I will still go ahead and have a heap of children and half them could have Huntington's – I had one – 50-50 chance that she might have it, hope that she doesn't and deal with it if she has....but not carry on having them in the hope.....I wouldn't have been selfish like that.....*

There were participants who would not consider having children until they had been for predictive testing and found out they did not carry the HD gene. They were perhaps able to manage the uncertainty in relation to themselves but when they considered the possibility of having children they appeared to want certainty before deciding to start a family:

04: *Erm, we put off having childrenbefore I knew one way or the other*

The participants who managed the uncertainty over their HD status by trying to get on with as normal a life as possible appeared more able to ignore HD and not let it dominate their everyday lives, compared to those who searched for signs of the disease and almost lived as if waiting for the disease to start. Interestingly, there was one participant who appeared to manage his uncertainty by becoming more involved with HD through choice. Instead of trying to ignore the disease or become troubled by thoughts of HD the participant set up support groups, searched for information on the disease and raised money for HD charities:

06: It was blooming awkward on flag days and things – you shook a can in front of somebody and say I'm collecting for cancer that was fair enough or I'm collecting for ME or something, but if you said I'm collecting for Huntington's Chorea you got "what?" You spent as much time explaining and lost a fortune (laughs).....so yeah and then in 1989 by then there was I think another few support groups and the HD association suggested we set up on our own....which we did so yeah we set it up and took it from there so that is how we got involved in the international HD scene and that's how I met other people/families with HD. I mean I think being involved helped a lot.... I suppose you thought you were doing something eh.....for what help I had received, and you were you were involved and I was putting a wee bit back.....

Having already lost a sibling to HD perhaps inspired his involvement in HD campaigning and support groups. It is possible that for some people, finding out information on the disease and setting up support networks helps to manage the uncertainty associated with living at risk and gives a sense of control over the situation.

3.2.3 Making the Decision

This subcategory explains how the participants decided that they wanted to go for predictive testing. Although all of the participants went for testing there was variation

in terms of when they made the decision to be tested, whether they kept the decision a secret, and their motivation for testing. Initially some participants decided not to get tested after finding out about their risk of HD and instead lived with the uncertainty for a varying number of years:

03: And you see that's in a sense why I decided not to be tested the first time I thought of it because I thought you know if I have got this you know this is a point in our lives when I don't have to think about this so what is the point in bringing to the fore... uh I got tested when I was 36, no that's not right, when I was 46 – 30 years after finding out, so I knew I was at risk for 30 years.

This appears as though participant 03 wanted to try to forget about HD as she was at a stage in her life whereby she had her own family and was living far away from her father who had HD and did not have to see the impact of HD on him and her mother. Initially some participants started the testing process in terms of going to speak to a Clinical Geneticist about predictive testing but decided not to go ahead. For example, the following participant decided not to get tested for similar reasons to participant 03 in that she was happy with how her life was at that stage and perhaps did not want to risk changing this:

01: I'd been back and fore to see Dr X a few years before that...and eh she asked if I was wanting the test and I says no, I was happy how I was. I had my husband and my two boys and I was fine....

Other reasons for not going for the test included the situation whereby a spouse was not in favour of her at-risk husband going for testing:

02: and em we spoke about taking the test...we went up to see Dr X a few times and erm my wife she wasn't too keen.....so we left it a couple of years...

The nature of HD as a disease means that it is something that can be put to one side for a while until the time comes that it needs to be addressed. Although the above participants knew they were at risk of the disease it wasn't immediately affecting them and even if they did have HD then the symptoms may not start for another 10 or 20 years. As a result it appeared to be preferable for some participants to live life as normally as possible and not disrupt the present situation until something happened that caused the risk status to become an issue again. One of the main reasons people changed their minds and made the decision to be tested related to either a desire to have children or a need to remove the uncertainty for children that were already at risk:

09: When I went for my first scan it was the first time I met Dr X.....erm and we spoke about me getting tests done on the baby and myself, erm and they could also offer me a termination if I wanted it knowing that the baby had it, and I just went – I seen my baby for the first time a couple of hours ago and you are speaking about terminations and I said no so Dr X says go away and have a think about the test....because they were starting to pick people for the first testing.... and I said yes I wanted to do it, and she said no wait until after you have had your baby and then get in contact with me.....so I waited until my eldest daughter was born – and informed her that my daughter was born and that I was still interested in going forward so I went back and saw Dr X

The above participant decided to go for predictive testing after having her first child as she didn't want to put any further children at risk of the disease. Other participants made the decision to be tested when their children had grown up and were either starting relationships or were getting married and were thinking of starting a family:

03: I came back to see Dr X and I mean....the way she does it is....I felt is that is sort of took me on some sort of a journey almost – that I was wondering if I wanted to do this – by the time I got to the point of blood tests you know I had moved from where I was to a place where I really was sure that I wanted to know the results and erm.....I mean...the issue to me was that if I didn't have this disease I didn't want to spend the rest of my life

worrying about it and I didn't want my kids to have to you know spend their lives worrying about it. My oldest son by this point was actually 16.....I mean he hadn't got into any serious relationships at that point and I thought do I want to burden him with this concept that he can't have a girlfriend without telling her about this you know which is actually a fairly horrendous thing to do to anybody, and I mean to follow my mum's edict you know you've got to tell somebody before they are serious about you, when you don't want to tell somebody that sort of thing until you are serious – you know what I mean.... you've got to be certain within yourself that you want to know this for yourself.....but erm I mean that's largely why I got tested when I did was that I just felt that if my kids didn't have to think about this, it was just – I didn't.....if I could take that burden away, if I could just get rid of it I might as well do it you know.....

Like the above participants most of them who had children explained that they were primarily taking the test for their children but also to remove their own uncertainty:

01: *We went back and forth and spoke to her (Doctor) about things and then one of the times we came out X (son) turned to me and said you dinnae have to do this for me and I said yes I do.....I need to do it for myself and my sons.....and my husband...*

10: *When I knew the risk I didn't really, I wanted to know first myself before I delved into it with my sons. I didn't see the point in putting them under pressure unnecessarily so I just took it on my own shoulders until I got tested.*

For some participants, it appeared as though they became more anxious as their children approached adulthood that they may have passed on the HD gene to them:

02: *No, no....it was mainly for the kids I went, because the children they were getting married at the time as well so eh....I just decided to.... I mean that was the worst thing as well, passing it on to the children and I didn't want that to obviously happen...*

The following participant was in the difficult situation that when he found out he was at risk his daughter was already pregnant with his first grandchild which meant that they could both potentially have the disease:

08: *my daughter at the time, she was about 18 or thereabouts. Unfortunately she was in the situation where she had fallen pregnant and you know.....we wanted to know, basically I wanted to know because of my granddaughter.....and I wanted to know then*

basically if I had carried the HD gene....and my eldest lad, he was married at the time, but no family and it was really the reason why I wanted to find out, you know if I carried the faulty gene was specifically because not for myself but for the family....

There were a few participants in the study who for various reasons were unable to have children so their reasons for taking the test differed to the rest of the participants. The following participant found out he was at risk at a time when testing was not available and he had to wait a number of years before he had the opportunity to discover if he had the HD gene:

06: The impetus was really the discovery of the gene and the more reliable test....

Participant 07 made the decision to be tested as his brother was also going for testing so they went through the process together:

07: My mother was tested and she found out she had it and then erm we was all tested at the same time, we just all decided to get tested at the same time....it came from my granny you see....

When making the decision to be tested some participants went through a process of thinking through the consequences of both favourable and unfavourable results. Although they would have possibly had to do this with a psychiatrist as part of their assessment to see if they were suitable for testing some were more willing to consider both scenarios than others. The following participant appeared to have thought about how she would cope with either result:

03: To get tested would be a way of um you know either I haven't got it, we don't need to worry about it or if I have got it you know at least I know that I really do have it but

the issue there was very much I felt was I had to almost kind of convince myself – I really do want to know if I've got this which is actually – of course you don't really want to know if you've got it.....but you know.....if that result comes back that I've got it then I have to be sure that I have made the right decision to have this test and and erm you know I think I felt that it would be the right decision because at least I would know that there really was something to worry about rather than me spending the next.....

Whereas other participants appeared to try to put thoughts of an unfavourable result out of their mind:

01: I always tried to be positive, and try to put it to the back of your mind, it wouldn't always go but I just had to try and be positive....

One participant went ahead with testing without informing his wife and children as he knew his wife was not in favour of testing and he did not want to worry or upset her:

02: Had I got the wrong result I don't know what would have happened...I wasn't prepared, I didn't know whether I would tell my wife or when I would tell her....but I mean I would have to tell her obviously...

In relation to this, it was evident that he had only really thought about the consequences of a favourable result and hadn't considered what would have happened if he had found out he had the HD gene.

3.3 Trying to Distance Self from HD

The participants' journey into the unknown ended at the point they received their favourable result from predictive testing for HD. *Trying to distance self from HD* is the second main category and it explains the process which participants then went through as they attempted to adjust to their new genetic status. The participants appeared to vary in how successful they were at distancing themselves from HD and this at times depended on whether they had emotional or geographical distance from the disease.

The subcategories; *facing the truth, breaking the news and moving forward* explain what happened to the participants when they received their favourable result and illustrates the variation in experiences and success in trying to distance their selves from HD.

3.3.1 Facing the Truth

The subcategory, *facing the truth* describes the moment when the waiting is over and the participants receive their test result which marks the end point in their journey into the unknown. The participants lose their at-risk status and the uncertainty over their future health is removed. The subcategory highlights how many of the participants had prior predictions or hunches about the eventual test result and outlines the emotional reactions experienced by the participants on hearing the news that they were no longer at risk of HD. A few participants stated that they did not think they carried the HD gene and had a feeling that they were going to get a ‘good’ result:

08: *I think you know in reality you think to yourself....personally I always felt very positive through it.....I think at one point X (Geneticist) asked me at one point how do you feel personally.....and I said well to be quite honest with you I feel okay, I don't feel negative about the situation, as a matter of fact I think maybe I said to her I don't carry the gene, you know the faulty gene*

04: *Yes, I thought that I was going to be okay and that I didn't have it....I just had an idea, a feeling that I didn't have it....*

06: *I don't know I just believed that I didn't have it. There was nothing to base it on....*

None of the above participants were able to give reasons for why they thought they did not carry the HD gene and basically described the prediction as a “feeling” that they

had. In contrast, there were some participants who had the feeling that they were going to get an unfavourable result:

05: It would be pretty lucky if all three of us (siblings) had been negative. I was pretty sure one of us wouldn't be. Yeah and I think in a way I was pretty sure it would be me because my great-aunt who we probably think had Huntington's – my mum had always said that I was very, very similar to her..

The above participant appeared to base her feeling on the fact she resembled a relative who had HD and therefore thought she would be more likely to get the disease. The following participant had no reason for why she thought she would have the HD gene but as soon as she found out she was at risk it appeared as though she expected to have the disease:

10: I immediately thought I have to know because if I knew I would have to start organising and telling them (sons) and explaining to them and see if they were willing to get tested.

When the participants received their test result, their emotional reactions ranged from joy and happiness to feelings of indifference. A common initial emotion was that of relief. Participant 03 explained some of the fears and worries she had about HD when living at risk and how she feels relieved as she doesn't have the same anxiety about the future:

03: I think I probably did cry but I didn't cry.....you know it was just such an overwhelming relief – for yourself and for your kids and.....for X (husband) as well – you know it just.....such a huge of burden of anxiety and fear for the future and the sense that you don't have a future as well – you know the sense that well even if I don't have it now, you know there is not going to be, I'm never going to be elderly, an older person because by that stage I won't be fully functioning you know – so that doesn't exist for me....

Some participants who had children appeared to experience greater relief for their children than for themselves:

01: Just sheer relief....for my boys, for them and as I say the tears were flowing – I was hugging her (Doctor), I was hugging my husband. It was just sheer relief

The following two quotes illustrate the emotional intensity of the actual moment of receiving the test result, and the impact this has not only on the at-risk person but the family too. Participant 09 and her father witnessed the deterioration and death of her mother and the test result brought enormous relief that her child would not have to experience this and also her father would not have to see his daughter with HD:

09: My thing was I was only interested in getting a result for my daughter for knowing my daughter was going to be fine....erm when it came to getting the actual test results I took my father with me and I also took my daughter. She was only about one and a half.....and Dr X I remember opened the door and she looked at me and stood there with a smile and you still had the chance to refuse getting told when you went in, and she just looked and said I'm not even going to ask if you want to know or not and that's because it is good news you don't have it. And I just sort of sat there. I felt so relieved for my daughter, not for me and the first thing I turned round to my dad and says was – I can have heaps of kids now (laughs). And after that I think my dad was sitting crying and I was just sort of more gutted (for brother), nae gutted I was totally chuffed to bits because my daughter wasn't going to see me like I seen my mum.

Similarly, participant 10 had experienced the impact of HD on her sister and her father and was therefore extremely anxious about receiving her test result. Despite receiving a favourable result and feeling relieved, she is still struggling to forget this day:

10: Oh it was just awful – it was the worst day of my life. It had been leading up to it, and watching the calendar and....by this time I didn't ken what I thought. There was so

many things going through my mind, trying to imagine that I was okay and if I wasn't how am I going to tell the boys.....what is going to happen, just horrendous.....I've never been right since.....I still remember the day. We sat in the waiting room and my friend was great, she was trying to make jokes and I felt so anxious. And Dr X eventually came through but it was like she had a mask on her face so you can't tell and my friend looked at me as we thought Dr X looked a bit solemn. We went through and we sat there and see when she told me – I got up and hugged her and she said you better hug your friend. It was relief....because I had seen before this time what Huntington's did to people through my dad and my sister

Although some participants felt relieved this was marred by the fact that siblings were still at risk:

05: Yes relief, we booked a lovely holiday away and we came back and started a family but it is very short-lived because of the impact on the rest of your family. When I was negative I thought well that's it, it is going to have to be my sister. It doesn't make sense at all but it is just the way I felt.

There were some participants who did not report feeling relieved when they received their favourable result. Participant 07 went through the testing process with his brother and they both received their results on the same day. He experienced a mixture of emotions in that he was pleased he didn't have HD but he felt upset for his brother and his mother. Participant 07's situation was unique in that he was born with a physical disability and as a result had hoped he would have the gene and not his brother:

07: I found it more difficult when I found out I didn't have it and knowing that my brother had it. It is a favourable result but it's nae good because somebody in your family has still got it and is still coping with it.... I was happy with it but as I say I was upset for my brother and my mother but I mean there was nothing I could do anyway you see.....it was out with my control. If I could reverse the results I would you know..... I was hoping it was the other way round for my brother because I'm in a wheelchair anyway.... it wouldn't affect me so bad because I thought you see because I was in a chair anyway....

The following participant described experiencing very little emotion on hearing his favourable result and suggested that perhaps this was because the news was what he predicted:

06: Dr X said – you know I'm kind of worried about you, you just stood there and took it and never said a word you know....my wife showed more emotion. I mean I think I slightly worried myself as well but.....I keep thinking that was the answer I expected – if it had been the other way then I don't know to be honest.....well you cannae theorise about what never happens..... I don't know I just believed that I didn't have it....

Similarly, participant 08 reported feeling indifferent after receiving his result and it is possible that this is because he also had predicted that he did not have the HD gene:

08: Anyway X eventually came out and took us in and she was sitting at her desk and then basically just stood up and put her hand out and said congratulations she says you are not a carrier of the faulty gene. We sat there and we had a little bit of a chat....and she was saying how do you feel about the negative result, you don't carry the faulty gene.....erm and I said I don't feel up nor down....you know I really didn't feel up nor down....I went in to it with a totally open mind about everything and you know I sat there and took the information and whether X was looking for some sort of elation or what I'm not quite sure.....Erm but no I didn't feel up, I didn't feel down, I was on an even keel you know

3.3.2 Breaking the news

After the initial relief experienced when receiving the favourable result participants then had to face the task of *breaking the news* to other family members. In normal circumstances news that someone has been spared of a terminal disease would be celebrated with other family and friends; however, the situation is different with HD. The participants were usually able to share the joy of the news with immediate family such as spouse and children however it was communicating the result to the wider

family that was more complicated especially if a parent had the disease or siblings had HD or were still at risk. This subcategory highlights the range of experiences and associated emotions when the participants decided to break the news of their favourable result to their family. For the participants who were married or had partners then most of them had also attended the clinic to hear the result. However, there was one participant who kept the testing a secret from his wife as he knew she was against it and he did not want to worry her. He told her the news when he got home from the clinic:

02: I bought a bottle of champagne at Tesco and stuck it in the fridge and when I told her.....I told my wife as soon as I got in. She was over the moon, particularly for the kids because eh that was the main thing, passing it on to the kids and that...and when the kids came home I told them as well....because they, they were looking HD up on the internet and all that so they were, they obviously feared the worst because they knew my father had died and he had it.....because we never kept nothing from them just in case they did get it...so, and then they were obviously over the moon...

In terms of immediate family, the participants who had children were keen to share their favourable result with them as it had positive implications for their lives and future plans:

03: So erm, we went home and my husband bought a bottle of champagne and my oldest son said – my husband had called them in for tea and they saw we were having a bit of a celebration and X (son) said why have we won the lottery and X (husband) says no better than that.....and in fact (son) guessed what it was – I mean he was sufficiently tuned into the issue to get what it was – and I think it has made a huge difference to his life in that erm....you know, well it's made a huge difference to all their lives obviously but I mean he was the one who it was weighing down on most and you know therefore knowing gave him you know, released him from that burden.....

Being able to release the burden of worry from their children seemed to be a huge relief for the participants.

01: X phoned me, he was at work at the time – on the Thursday evening, and I just said it is good news X, and he says I knew it mum, I just had a good feeling all day. I told him that he would be getting a letter from Dr X for him to keep, saying that everything is fine and then I phoned our son in X and told him and he was just like – he is more laid-back, he doesn't get himself worked up about things as much as (other son) but he was still pleased, ken fit I mean, happy...

The following participant proposes that the lack of reaction from his children to the news could be due to their age:

08: But even afterwards when I got the favourable result and that and I told them – there was no elation or “that's great”, it was more like “fine”.....but you know when I mentioned to them that I was going for predictive test, I did tell them and they never really wanted to know, well when they said they didn't want to know erm they weren't that interested, they weren't concerned lets put it that way..... whether it is youth on their side I don't know, but this you know “we are young” attitude you know so – a bit pensions like you know – im 25 I don't need a pension yet.....

Generally the reactions of the immediate family to the news were positive and some participants described celebrating their favourable result. Usually the participants broke the news to the immediate family first and then afterwards they decided to tell siblings, parents and the wider family. This seemed to be more difficult for the participants who had family members with HD or still at risk of the disease. The responses of the siblings and parents to the news were also positive but it appeared to be the participants who struggled with telling the news:

01: but eh the hardest thing was going in and seeing my auntie in X, my uncle's wife because their two daughters have it... and it was hard going to tell her although she was really good, she was really good.... (sighs), she was.....maybe relieved for me but yet I felt.....sorry for her, for my two cousins.....for me to go and tell her that I had been tested and I was fine.....that was the hardest thing...

There was a sense from the participants that prior to receiving the result they shared a common bond with the rest of their at-risk family. However, the favourable result changed this and made them in some ways different to their family members:

03: It is a terribly difficult thing to tell members of your family who are still at risk that you know "I'm okay", "you're still.....", and there's this thing you know – the concept that you are all in the same boat and suddenly you've bailed out and you are not in it anymore and, and I don't know if there is any right way of doing it actually, there probably isn't a right way of saying actually I haven't got it – I've found out I haven't got it I'm okay.....um....Well she took it you know very positively and fine and it was, but you know I think it is fairly devastating but, sorry I mean that makes her sound awful, it's not like that at all you know....it's the two – of course you are very pleased that, she was very pleased that I hadn't got it but at the same time you know it is quite hard.....

Participant 03 implied that her at-risk sister possibly hid her true feelings when she heard the news, and that although she was pleased about the favourable result it may have been upsetting too as she may have thought that it meant she was more likely to have HD. The following participant hoped that her favourable result would encourage her brother to go for testing but he would not change his mind:

09: I went home and told my brother and he was chuffed and I says to him look will you still not entertain it (going for testing) and he says no.

02: then we went and told my sister and my brother and I explained that to my mum of course....they were happy for me mostly and they decided....well, they were okay not knowing because they were worried about insurance.....they were taking on insurance policies at the time so....but maybe my brother, he will probably go along one day.....I imagine he will take it....

For the participants who had siblings in the advanced stages of HD only one of them decided to try to explain the result to his sibling and the others decided against it as they

were either unclear as to whether the news would be understood or did not feel it was necessary to pass on the information:

08: *I don't think, I mean he could have probably taken that in and probably realised but again there was no emotion or back slapping saying that's great news, at least you won't have the problems that I am going to experience.....*

10: *No, by that time she was really unwell, no. I never ever said to my sister about Huntington's. I never had the chance to speak to her about it. That's the thing about Huntington's – I'm nae sure how much they understand?*

04: *No, I didn't think he needed to know....*

3.3.3 Moving Forward

Once the participants had received their result and had broken the news to their family they appeared to try to move forward with their lives and distance themselves from HD. The subcategory *moving forward* explains how it seemed to be easier for some participants than others to get on with life after their favourable result, and it appeared to help if the participants had either *emotional* or *geographical distance* from family members with HD. There was only one participant who appeared to move forward with his life very quickly after receiving his test result and he described being able to forget about HD:

02: *We are fortunate that way that I've got a good job and that eh....we just always carried on and enjoying life as normal.... we can ignore Huntington's now...maybe it's whether I shut it out of my mind or whatever, just get on with it – that's the kind of thing I put to the back of my mind and then never think about it, I never really think about it until I get things like this (research).....but we never really speak about it. I mean, I'm not really an emotional person really – I think eh I just seemed to get on with it, I'm just really laid back....that's what I think, that seems to work*

This participant seemed to attribute his ability to carry on as normal after the result to his laid-back personality style and approach to life. He had a relatively positive past experience of HD and therefore didn't appear to be concerned about his at-risk siblings:

02: I never really think about it....well they never show any signs of it, my sister and brother....

Furthermore he seemed to be able to get on and enjoy life and although he thinks a cousin may have the disease he is not emotionally or geographically close to her and is able to put HD out of his mind:

02: I'm sure her (auntie) daughter has got it eh....and as I said we never really see her, we're never, we actually forgot about it, speaking about it just reminded me....

The following participant also adjusted relatively well after her favourable result and she appeared to be trying to make the most of life now that she did not have to worry about her future in terms of HD:

01: My husband and I are doing a lot more things now, going on a lot more holidays whereas if I had HD.....then I couldn't have done that with him. As I said you can look forward to doing more things, make the most of it, you're fine now you ken you haven't got to worry about it....

Although all of the participants were pleased not to have the HD gene there were a few who described initially feeling as though they had lost something:

06: In a funny way I remember saying to Dr X that it was like losing something because it was almost like you had been going around for X amount of years with a chip on your shoulder and then all of a sudden someone had taken it off.....or you had been carrying

a heavy knapsack or something and you had carried it 24 hours per day and then all of a sudden somebody took it off – you know where has that gone?

03: It is very much part of who you are the fact that you have got this risk which is, I mean in thinking about coming here today I mean I think that was the main thing for me was that being a person who is at risk of having had Huntington's is actually a massive part of your identity, you know this is the burden who I am, and then that is suddenly taken away and it's actually quite "oh well who am I then?" And I mean it sounds pathetic because in a sense, why would you want that as part of your identity – of course you don't want it but nevertheless it is.... I was thinking what happened to that fact that I felt I had lost that bit of my identity and somewhere along the line between eight years ago and now it's become irrelevant – it is not a troubling thing in anyway.....

Both participants had lived with the risk of HD for a long time which possibly explains why they felt almost at a loss when they received their result. The risk was something they were used to and it in some ways it defined themselves and their wider family. Participant 03 explained, however, that with time this feeling disappeared. Another difficulty for this participant in adjusting to her favourable result was feeling that the risk had somehow protected her from other diseases and therefore she was now more likely to get another disease:

03: The other thing about getting a favourable result it that suddenly this particular thing which is the nasty thing in life which is going to get you is removed which means that actually you can then be got by all the other nasty things in life but of course this is totally illogical – people can die of heart attacks who have got Huntington's or they can you know they can walk under a bus like anybody else can but somewhere in my mind was this concept this is my nasty so therefore I'm not going to get the other nasties.....so you then in a sense feel more vulnerable....

Mental health problems were experienced by one participant a few months after his favourable result. Although he was unsure what caused this to happen, he suspected it

was in part due to seeing his brother with HD deteriorate whilst knowing that he had been spared of the disease:

08: It was a couple of months after that (getting the result), that I had a breakdown, I just freaked out....I was sitting in front of my computer....I don't remember it, my wife came down stairs and I don't know how long I had been sitting there staring at the screen.....I had just gone, what caused it we don't know.....I did erm have consultations with a psychiatrist and his team.....and I actually attended psychiatric hospital.....I'm actually still on anti-depressants now and erm we couldn't come to any firm conclusions about what had caused the breakdown um there was a lot of things happening, problems at work you know, this thing with my brother as well.....it was just like a knock-on-effect, several things all come together at the one time, you know something gave, and my body just shut down and said you need a rest. I lost about two months. I off work for about 2 months while I got back on track to recovery.....

The initial stages in moving forward after the favourable result were therefore more difficult for some participants than others. All of the participants were trying to get on with life after their result. However, HD still had an impact on most of them ranging from those who found it difficult to switch off from thinking about the disease to those who were actively caring for relatives. Although participant 01 appeared to be able to move forward with her life she still thought about HD a great deal. Her cousins have the disease and although she does not see them regularly she appeared to be emotionally close to her auntie and often thought about their lives and what may happen to them:

01: And I keep thinking, as I said my mother and my uncle died at 54/55 – how much longer have they (cousins) got – X is I think.....51/52, and X is 3 or 4 years younger than that, how much longer have they got? I keep thinking about them. My auntie is in her seventies now – is she going to have to bury her two daughters, that's what I feel, I feel for her....

Participant 06 appeared to be able to move forward without thinking too much about HD apart from when he hears of the death of friends or acquaintances from HD. Both of his siblings died a relatively long time ago and although his nieces have the disease he is not emotionally close to them because after his sister died, his brother-in-law found a new partner and they did not have much contact thereafter.

06: I don't think an awful lot about it.... I mean okay I don't have to worry about the children – it comes to as I say – a friend with HD died earlier this year so it sort of brings it back a bit you know and you hear about other people passing on you know.....I mean for me personally no, it's not so much to the fore as it was....

If a sibling has HD then understandably it is more difficult to ignore it and move on with life. The following participant explained that the disease was still in her family's life due to her brother having the gene and being symptomatic:

03: Well you see if my brother didn't have it, he wouldn't have it, it would be a dim and distant thought but because he has got it, us as a family, it is still very much pertinent. You know I listen to these messages from my brother everyday you know I use my mobile to phone another mobile to pick up the messages and I've got to go to X for a meeting but anyway I'm going to go and see my mum and I'm planning to go and visit my brother you know while I'm down there which is if he was well, personally I wouldn't do. I would almost certainly see him once a year if that you know so I mean in some ways the fact that he has got it and I haven't has definitely made us closer in the sense that I pay far more attention to him than I possibly would have in other circumstances.....

On a more positive note participant 03 acknowledged that receiving the favourable result has brought her closer to her brother as she visits him more regularly than she did before they knew he had HD. Although participant 03 had geographical distance from her brother and wasn't seeing the everyday struggle of HD, she received daily phone messages from him which made it impossible to ignore the disease and its effects. The

following participant reported that as time goes on coping with HD is more difficult as she has to witness her mother and siblings deteriorate:

05: I would say that it is certainly harder.....I think really because now I look at mum and I think – what is going to happen to my brother and sister. Until you see what Huntington's can do it is very difficult to visualise, you read about it but until it happens to someone you know you can't see how it impacts. I had a wobbly probably about 18 months ago and the area HD advisor who managed mum's care at the time referred me to a counsellor and I went to see her. By the time I got an appointment I was absolutely fine. It was just really as mum was going in permanently and I was really struggling with it.

Participants who had siblings with HD sometimes questioned why their brother or sister got the disease and not them and feelings of guilt were reported. The following participant struggled with the fact his brother is in a nursing home and is not the fit and healthy person he was previously:

08: I mean obviously my brother has HD and it does concern me. When I see him, I will be honest with you, I think every time I see him I sit there and the same old question goes through my head – “why him”?, and why not me? It really does and to see him – he is.....pathetic, you know and he is in a situation where he sits mainly in his room at the nursing home, he has no interaction really with other people. But I do go down as often as I can but what I have been noticing over the last wee while, whether it is because he is on his own a lot more erm his speech is starting to deteriorate quite badly to the extent now that you know it is very, very hard to have conversations, and again a negative side and a guilt trip comes in because you can't really spend any more than an hour or 45 minutes with him – you are really stretching it at an hour because you get to the stage where you think I've got to get out of this, I've got to get out.....

Even after the death of a parent or sibling to HD it appeared to be difficult for some participants to move on with life and distance themselves from HD. Although life had new opportunities after the death, such as working full-time and having more time for themselves, the emotional impact of being in a family with HD was still evident. For

one participant who lost her mother to HD at a young age and then after receiving her favourable result had to care for her brother with HD until he died, she at times still experienced problems as a result of her past:

09: I couldn't deal with him (partner) and I know it sounds bad but I just wanted to push him away – because it was like – you are going to die anyway – and he was like – what – and I says you are going to be like the rest of them – and he says everybody dies and I says – I know but if I get rid of you now I don't have to deal with it later so erm i broke up with him and he couldn't understand it and I said I just need time for me.....even getting up in the mornings, some days you think god I can't be bothered the day and then you think how can you nae be bothered, there is nothing wrong with you – what is your excuse for not doing something. Nearly every day I take my dogs for a walk and I go walking and I stand there and think – how lucky am I – and other people they just take everything in their stride – (gets upset), sorry....

Similarly, the following participant did not find that life became easier with the passing of time and was still deeply affected by the death of her sister from HD:

10: But at the time I was probably happy (getting the result) but looking back now I think (sighs), it was like a false happiness really...because you were happy at the time but as the weeks and the months went past and you really had time to think about it, the reality of the situation begin to sort of settle in.....But I wouldn't say that as time goes on you feel better, because you dinna it is something that will be with me until the day I die.....

There appeared to be the issue of *divided loyalties* for participants who described trying to live a normal life with their immediate family but at the same time feeling the need to care for siblings or a parent with HD:

05: Sometimes he (step-dad) makes me feel guilty for not visiting mum and I should or he thinks I should but if you are working full-time and you've got 2 kids to look after. We never used to go and visit mum every single day – we have got our own lives. He is her husband at the end of the day and that kind of puts it into perspective and takes some of the guilt away but you do feel or I feel like I should be down visiting but it

means taking the children and they don't want to sit across from granny and see other people who are far worse as well. It is not a nice day out....

The participants appeared to want to protect their children from seeing the devastating impact of HD but also wanted to care for their family members:

09: Erm I just, I didn't, I just took one day at a time. Because I had three young children, they just accepted it as part of their life as well....they all knew about Huntington's – they all knew what it was, they all knew if he threw a tantrum it wasn't aimed at them. It was hard for them to see, and it was hard for me when my brother lost the head with my kids because that's one thing with me – you can throw and shout and scream as much as you liked at me but don't do it to ma kids

3.4 Having a Sense of Duty

This category explains why some participants struggled to distance themselves from HD. *Having a sense of duty* to family members with HD interfered with some of the participants' ability to move forward, get on with life and ignore the disease. A sense of duty was demonstrated by participants not only by caring for family members with HD but also through fighting for their rights and protecting them and giving something back in terms of doing charity work. Some participants chose to get involved with HD groups and charity work when they could have tried to forget about the disease after receiving the favourable result. Having a sense of duty also meant that through caring for family members in the advanced stages of HD some participants had to witness their deterioration. As a result HD was still very much a part of their lives.

3.4.1 Caring

Despite living a long distance from her brother the following participant still arranged his food shopping online and also was in daily phone contact with him to take the pressures of his daughters:

03: My feeling is that this is the price I pay for not having it myself that you know I can, because you know I've said a couple of times I'm not close to this person – I haven't been close to him my entire adult life but nevertheless I kind of owe it to him if you see what I mean – It's kind of I haven't got this thing, my kids haven't got it you know it is the least I can do to try and you know make things a little bit better for those who have or for those who are still at risk.....

The high levels of stress associated with caring for a person with HD was clear from the stories of some participants. However, despite this they continued to care for their family members until their death. The following participant was the main carer for her brother and had to cope with the behavioural and personality changes associated with HD. On one occasion she was physically attacked by her brother but still continued to care for him despite her anxiety:

09: No, I had a rather sore head, and still do so it is my own fault for not getting it checked out at the time....I was in a total daze. And that was the first time I felt I could turn round and say I hated my brother but I knew it wasn't him but still I hated him, one – for lifting his hand to my dad because my dad wasn't able to hit back, and two – for taking it out on me for accusing me of something I didn't do, and I thought what am I doing, I've got my own family, I dinna need this.....erm...I was actually sweating being around him....and if he shouted for something I was like (tenses up), and I thought that's nae me, I can deal with this, why let the disease get the better of me, I says no that's not the way, so I had to take a step back and pull myself together and say when you go into your dad's you act normal even though underneath you are a bitty nervous....just be your usual happy self and I did and it sort of made my brother relax a bit more.

Having a parent and siblings with HD resulted in the following participant having to do most of the caring duties for her mother as she did not feel she could ask her brother and sister to help given they knew they were going to develop the disease:

05: I've done an awful lot of my mum's caring but she is in a specialist unit now. I did a lot to get the care package before she went in and I get narked at my sister sometimes because it always felt like it was on my shoulders then I feel guilty for feeling narked at her and you know sometimes – my sister is only an hour and a half away and I feel cross at her that she didn't come down to support me for a bit because I felt any day off I had I was down there.

3.4.2 Protecting

A few participants demonstrated their sense of duty through fighting for the rights of their relatives with HD. In both of the following examples there was a perceived injustice whereby the sibling with HD had been wrongly arrested for alleged drunken behaviour and also unfairly treated by an advisor at the job centre:

08: There was actually a letter that came in from the procurator fiscal saying that this time nothing would happen about it and if it happened again he would be charged and taken to court.....I was not very sympathetic with them and I actually picked up the phone and I phoned this guy and demanded to know – why was my brother charged with this in the first place....and he said oh well we didn't know, we didn't think – I said do you know this guy is ill – oh how would we know that.....I says well obviously someone thought he was drunk because the police were called you know. I says my brother is not a drunk, I says he never ever did really drink, he would have the odd pint....I says but he hasn't drunk for years, he has a condition called Huntington's disease and I tried to explain to this guy what it was but he wasn't interested.....didn't want to know, and I says well in future I says I think your constabulary and all the rest of it should be trained in you know these kind of situations.

09: And I said don't you ever speak to any human being like that again unless you want to be wiped off my shoe – and my brother was in bits because he thought this isn't right you are my little sister.....it was like me sticking up for him, and I says well you stuck up

for me all my life....I says payback time I will stick up for you, course I will you are my brother.....and it was hard for him to sort of step back a bit and let me nae take over but stick up for him because he was a male and he had his pride and all the rest of it, and that was the way he was brought up....a man is the man of the house and a woman is just eh second in command...

Fighting for their family members with HD was perhaps a way for them to feel that they were able to help and give something back. It was in some ways as if they were trying to compensate for not having the disease themselves and also being able to express their anger at the unfairness of the disease.

3.4.3 Giving

The following participants became involved in charity work for HD and in setting up local support groups for families with HD. This appeared to make them feel as though they were doing something to help others less fortunate than themselves and perhaps was a way of coping with the fact they received a favourable result unlike their siblings:

06: No, no I suppose it (the favourable result) reinforced it if anything because you know I had been there and done that and had the test, got the t-shirt and all the rest of it so I felt that eh you know I should be putting something back and you know there was an awful lot of people worse off than I was....

10: When my sister was living I got involved in a lot of charity work. I did a lot of car boot sales, did a sixties night and raised £3500 for Huntington's. So I don't feel guilty that I haven't done my bit for them.....you feel responsible for all the people.

3.5 Chapter Summary

In summary, the substantive model presented in this chapter highlights the processes which participants go through from the moment they find out they are at risk of HD through to a number of years after their favourable result. It illustrates the similarities

and differences between the participants' experiences which adds explanatory power to the model. For the participants in this study it appeared as though receiving a favourable result from predictive testing for HD was to some extent a relief. However, this was often complicated by the fact that the participants were part of a wider family system in which HD was still present. Many participants found it difficult to distance their selves from the disease after receiving the favourable result and for some HD was still part of their lives many years later.

Chapter 4

Discussion

4.0 Introduction

The aim of this study was to explore the experiences of people who received a favourable result from predictive testing for HD and how this impacted on their relationships in the long-term. The previous chapter presented the findings of the study which illustrated two main processes which the participants went through, firstly, *facing a journey into the unknown* when they found out they were at risk of HD and secondly, *trying to distance self from HD* after they received their favourable result. The findings suggested that although all of the participants found out they did not carry the HD gene most of them still carried the burden of HD to varying extents. This was mainly because of other family members having the disease, which impacted on them emotionally as well as in terms of *having a sense of duty* to care for them. After the death of family members from HD it appeared as though some participants struggled to forget about the disease and move forward, suggesting that the psychological impact of living in a family with HD may be difficult to overcome. This chapter will discuss the findings in relation to the literature on predictive testing for HD and also with reference to the Family Life Cycle Model (Carter and McGoldrick, 1989). The chapter will then explore whether the proposed substantive model has resonance for people with other genetic health conditions, such as hereditary breast cancer. The strengths and limitations of the present study will be discussed and the implications of the findings for clinical practice and research will also be presented.

4.1 Facing a Journey into the Unknown

There was considerable variation in terms of how and when the participants found out they were at risk of HD. Some knew from an early age as they were living with a parent with HD whereas others found out later in life when a sibling or parent developed HD in their fifties or sixties, and they had not been aware of HD in their family prior to this. Only one other study could be found which reported in detail how and when participants discovered their risk of HD. Etchegary (2006) described four different pathways to finding out; something is wrong, out of the blue, knowing but dismissing and growing up with HD, all of which were found in the present study. Also consistent with Etchegary (2006) was that most participants were ignorant about HD when they found out about their risk because there was an unclear family history of HD, with some relatives having been misdiagnosed in the past with multiple sclerosis or Parkinson's disease. The present study supported Etchegary's (2006) finding that participants wanted to speak about the past, present and future in terms of their genetic risk and overall genetic journey. Although the present study's intended focus was the time period after participants had received their favourable result it was quickly apparent that participants wanted to tell their story from when they found out about their genetic risk. Etchegary (2006) highlighted the importance of considering temporal and historical factors when working with individuals and families at risk of HD, and having an awareness of how a person finds out about their risk of HD, as this may influence how they cope with the testing process and final test result.

The substantive theory in the present study described how participants varied in their ability to manage the uncertainty associated with living at risk of HD. Some

participants lived at risk for many years before deciding to take the predictive test, whereas others decided very quickly that they wanted to be tested so therefore did not have to manage the uncertainty for very long. There were a few participants who appeared able not to think too much about HD and were managing to get on with their lives. However, there were some participants who searched for signs of the disease on themselves and were almost waiting for the disease to start.

Etchegary (2010) used a chronic risk perspective to explore the meaning and consequences of living at risk of HD. She used a qualitative approach and found that although for most participants the risk was chronic, it was not continually salient or a worry for them. The salience of the risk appeared to depend on zones of relevance which mean that there is considerable variation in the extent to which the risk of HD becomes relevant or prominent during a person's life (Etchegary, 2010). The stage in the life course and family history of HD were found to be the most significant zones of relevance and had a direct impact on risk salience. For example, those who found out about their risk of HD when they were young appeared able to get on with their lives without worrying too much about the disease at that stage. Similarly, the risk was less salient for those who were in their fifties or sixties as their chance of developing the illness had reduced (Etchegary, 2010). The risk was more salient for those who were at an age where they were considering marriage or having children or were approaching the age on onset of a parent with HD (Etchegary, 2010). From a clinical point of view, Etchegary (2010) proposes that zones of relevance are helpful in highlighting whether and at what stage a person at risk of HD may benefit from support.

Quaid *et al.* (2008) explored the experiences of people living at risk of HD and found that the participants carefully concealed their at-risk status from others and only told people that they trusted. Although a few participants in the present study decided against disclosing their status to specific people (local GP in small community) in general most participants appeared open about their risk and told partners, children and work colleagues. Only one participant decided not to tell her grown-up sons until she had received her test result and was certain about her own risk status. Quaid *et al.* (2008) also found that preserving hope was an important part of living at risk, and that the participants decided against testing mainly because of wanting to keep their hopes for the future alive. All of the participants in Quaid *et al.*'s (2008) study had chosen not to go for predictive testing and were living at risk at the time of the research, whereas the participants in the present study had all had the test and were reflecting back to the time when they lived at risk. In the present study, some participants appeared to have hope for the future in that they had a feeling that they did not have HD and even if they did have the disease, they did not think it would affect them until late adulthood, and would be relatively mild, as was the case with a parent who had the disease.

In Keenan *et al.*'s (2007) study some of the young at-risk participants who were growing up in families with HD, were described as the 'worried well' which is consistent with a few of the participants in the present study. They misinterpreted bodily signs as being symptoms of HD which made them even more anxious about their risk of the disease. The participants in Keenan *et al.*'s (2007) study who coped well with the at-risk status had good support networks and found out at an earlier age about their risk. Similarly in the present study the participants who discovered their risk as

children or teenagers appeared to cope better with the uncertainty, than some of the participants who found out later in life. The main reasons reported by the participants for deciding to go for predictive testing were family planning issues, or if they already had children, to remove the uncertainty for them. This is consistent with other studies into the motivation for predictive testing for HD (e.g. Evers-Kiebooms *et al.* 1989; Tibben *et al.* 1993a).

4.2 Trying to Distance Self from HD

The present study found that the participants who received favourable results for predictive testing for HD tried to distance themselves from HD, but varied in how successful they were in achieving this. Although there are many studies on the psychological consequences of predictive testing, only a few have focused specifically on people who go for testing and find out they do not carry the gene for HD. Williams *et al.* (2000a) found that people who receive favourable results from predictive testing go through a process of redefinition in relation to themselves, their family and their role in society when trying to cope with their new genetic status. The participants were more focused on themselves at one month post-result. However, at six months after the test they were starting to look to the future. Prior to receiving their test result most of the participants had feared the worst and had thought they would have the HD gene. As a result they were not prepared for the favourable result. This in contrast to the present study as many of the participants had a feeling that they did not carry the HD gene, and said they had not anticipated how they would have reacted to an unfavourable result.

Williams *et al.*'s (2000a) theme of paradoxical emotions was evident in the current study in that participants were very relieved and happy with their favourable result; however, they felt sad and anxious for their family members who were still at risk or who had the disease. Although the participants in the current study were aware that their relationships with at-risk or affected family members may change following the result, few expressed concerns about how they still fitted in with their families which was different to Williams *et al.*'s (2000a) findings. Only one participant in the current study described feeling like she had 'bailed out' and was 'not in the same boat' as her siblings after the test.

In terms of how the favourable result impacted on the individual's role in society Williams *et al.* (2000a) found that the participants wanted to appear normal and were relieved not to feel stigmatized anymore. None of the participants in the present study mentioned the stigma of HD. Feelings of guilt, however, were common to some participants in both studies in relation to being free of the disease compared to other family members. Also in keeping with Williams *et al.* (2000a) was the sense of wanting to help others with the disease and having a sense of duty to them.

Williams *et al.* (2000a) interviewed their participants at one and six months post-test result and they stated that the redefinition process was still ongoing at six months. Participants were still trying to redefine themselves, their relationships and roles in society. Although some had made plans for the future they had yet to follow them through. In the present study, some participants had gone on expensive holidays and were trying to make the most of life. A participant had started a new job after her

brother had passed away from HD as she felt more able to move forward with her plans then.

In the current study, a few participants mentioned that they struggled initially after the result in terms of finding a new identity. One participant stated that this was no longer a problem but that she could not pinpoint exactly when this changed. With reference to William *et al.*'s (2000a) redefinition process, it appears as though the participants in their study were still struggling with this at six months, but that the participant in the current study had completed the process of redefining herself eight years after the result. As with the present study it appeared as though the participants in William *et al.*'s (2000a) research were having difficulty distancing themselves from HD and stated that it was something that would always be with them.

4.3 Impact on Relationships

In terms of the impact of the favourable result on the participants' relationships in the long-term, the main relationship they talked about was the one with their siblings who had HD. Some participants reported that they felt closer to a sibling as a result of helping to care for the person. Prior to HD coming into their lives they had not been close and did not have much in common. They lived quite far from each other and only met up approximately once or twice a year. However, when HD became a problem for her brother she made the effort to visit him more regularly and even helped him at a distance arranging online shopping for him. Another participant reported that she felt she could not become angry with her sister because she had HD even though she was demonstrating minimal symptoms at that stage. The participant was spending a lot of

her time caring for her mother who had HD and felt unsupported by her sister who rarely helped out. The participant felt guilty for being annoyed at her sister but also understood that perhaps her sister found it difficult visiting her mother knowing that she would end up like her in the future.

The stresses of caring for a sibling in the advanced stages of the disease took its toll on one participant who admitted to feelings of hate towards her brother after he was physically aggressive to both her and her father. This demonstrates how difficult it is to care for a loved one with the disease but at the same time feeling the need to help because of receiving a favourable result. In the current study, one participant received a favourable result and ended up caring for her older brother with the disease and she was very protective of him. There were a few occasions whereby she stood up for him and fought for his rights. She explained that her brother found this difficult as he said it was his role to protect his younger sister and not the other way around.

Sobel and Cowan (2000b) found that predictive testing for HD had the biggest impact on three areas of family functioning; membership, communication and care-giving. The study was from a family perspective and interviewed whole families. When siblings received different test results, there were disconnections in relationships; however, when they received the same result the common bond was strengthened. This was not found in the present study in that most participants still appeared to have good relationships with their affected siblings, and for some participants their siblings did not get tested and were not symptomatic. Sobel and Cowan (2000b) discovered that those families who had a cohesive approach to testing were more open in their

communications with each other about the testing process, experienced less relationship problems and were less distressed by care giving issues than those participants who went through testing alone and didn't involve their family. Interestingly, three participants who received favourable results chose to disconnect themselves from their families with HD as they felt they had the right to after growing up with the disease and experiencing losses. This relates to the category of 'trying to distance self from HD' in the present study and it sounds as though these participants were able to do this successfully without feeling a sense of duty towards their family members. It appears as though they felt they had done enough already in relation to HD and that they deserved to move away from the disease.

A few participants in the present study reported that their marriages ended a number of years after receiving the favourable result. Although they did not say this was the reason for the separation, they explained that their partners had struggled to be supportive or understand the impact of HD on the participant and the wider family. This is consistent with Richards (2004) who found that it was the emotional impact of living with the risk of HD for many years that caused a few couples to end their relationship after receiving a favourable result rather than the result itself.

4.4. The Family Life Cycle

The family life cycle (Carter and McGoldrick, 1989) (see Chapter 1, pg. 31) is a useful theoretical framework in which to consider the findings of the present study. In summary, the model describes six stages of the family life cycle: leaving home/single adults, the joining of families through marriage/the new couple, families with young

children, families with adolescents, launching children and moving on, and families in later life (Cater and McGoldrick, 1989). The model proposes that stress in families is often high at transition points when progressing to the next stage of the family development process and that signs of stress are more apparent when there is a disruption to the developing family life cycle (Cater and McGoldrick, 1989).

In terms of the present study, the main category, facing a journey into the unknown, highlighted how participants were at various stages of the family life cycle when they discovered they were at risk of HD. Some participants found out at a young age and were at the 'leaving home/single adult' stage or 'the joining of families through marriage/the new couple' stage. Other participants discovered their risk when they were at the 'families with adolescents' stage or the 'launching children and moving on' stage and they therefore had the additional worry that their children or even grandchildren could also have the disease. Those who discovered their risk of HD at a young age appeared more able to manage the uncertainty of not knowing their genetic status and got on with life as planned. A few participants moved on to the 'joining of families through marriage' stage and had partners who were aware of their risk. For some participants they then progressed onto the 'families with young children' stage whilst still at risk of the disease. This caused tension in their wider families as there was disapproval that they were having children who could potentially have HD. They had to deal with the 'horizontal stressor' of having children and moving to the next stage of the life cycle and also the 'vertical stressor' or the threat of HD and the disapproval from previous generations of the family. Other participants did not make

the transition to becoming parents until they had been for predictive genetic testing and for one participant she decided against having children due to the risk.

The participants varied in terms of the stage in the family life cycle they were at when they made the decision to go for predictive testing. One participant was prompted to go for the test when her eldest child reached 16 years old and he was starting to show interest in relationships with girls. She wanted her son and other children to know their genetic status before they had to make important life decisions and progress to the next stage of the family life cycle. There were a number of participants who were at the stage of 'launching children and moving on' whereby their children were adults and were starting to have their own family. The participants wanted to have the test so that they could inform their children if necessary before they became pregnant. For some participants the normal development of the family life cycle was disrupted not only by the risk of HD but also by other factors such as disability and being unable to have children naturally.

Receiving the favourable result was met with relief by most participants and for some it allowed them to progress to the next stage of the family life cycle, for example having children, or removing the burden from their grown-up children and giving them the opportunity to provide grand-children free of the risk of HD. The immediate family system's reaction to the news of the favourable result was positive; however, some participants found it difficult to communicate the result to the wider family such as parents, siblings and cousins who were affected by the disease or still at risk of it.

Participants in the study were caring for siblings and parents with the disease at a much earlier stage than would be expected in the typical family life cycle. There appeared to be a sense of duty from a number of participants to care, protect and give something back to siblings who were affected by HD because they themselves had been spared of the disease. This role, at times, directly impacted on the participants' immediate family because they still had their own children to care for. In addition, participants' parents affected by HD were less able to fulfil their grandparenting role as the disease progressed, and the participants had to deal with the loss of parents and siblings from as early a stage as 1 or 2 in the life cycle as opposed to the final stage. A few participants were, however, able to move forward with their lives after the favourable result and distance themselves from HD, particularly if they had emotional and geographical distance from affected or at risk family members. The favourable result enabled one participant to almost re-start her life again once her sibling had died of HD and she was no longer caring for him. Her children were less dependent by this stage and for the first time in years she was able to think of finding a job which is something that probably would have happened much earlier in the life cycle had she not lived in a family affected by HD.

4.5 Relation to other Genetic Diseases

Predictive genetic testing is available for a number of other genetic health conditions such as hereditary breast cancer and ovarian cancer (HBOC). The findings of the present study could have resonance for people who are at risk of HBOC and who receive favourable results from predictive testing. However, it should also be highlighted that there are some important differences between HD and HBOC which

could have implications for how people cope with the at-risk status and test results. For example, if someone has the HD gene then they will develop the disease; however, with HBOC if people have the gene mutation they are given a percentage risk of getting cancer and may never develop the disease (Hamann *et al.* 2008) Also if people are diagnosed with HBOC there are treatment options available which may either get rid of the tumours or keep the disease at bay for many years. With HD there is no cure, it is degenerative and the person can only be given medication to help control the symptoms (Hamann *et al.* 2008). From a family point of view both illnesses are devastating but with HD the affected person can change in personality, become cognitively impaired and display challenging behaviour all of which put an enormous strain on families.

Dancyger *et al.* (2010) compared the motivations and attitudes of family members towards genetic testing for hereditary breast and ovarian cancer. The motivations for testing were similar to that of the current study in that most participants stated that they wanted to take the test for others and less so for themselves. Hamann *et al.* (2008) discovered that siblings who received similar test results had less relationship difficulties than those who received different results. Dudok de Wit *et al.* (1998) found that the pattern of distress over time was similar for both carriers and non-carriers for HD, HBOC and familial adenomatous polyposis (FAP). They therefore proposed that predictive testing for HD could be used as a model for other genetic health conditions but also highlighted that those participants at risk of HD demonstrated higher levels of distress than participants at risk of the other diseases.

4.6 Strengths and Limitations of the Research

Although this study is small and exploratory, it adds to the body of knowledge on predictive testing for HD. There is a need in the literature for more studies which focus on individuals who receive favourable results from testing. This study therefore met this need and also included participants who were at least five years post-test result, which is important as there is a lack of research which explores the long-term psychological impact of predictive testing for HD. In addition, the study has developed a substantive model which hopefully will be further explored in future studies in this area. The substantive model has also helped to highlight the processes which individuals at-risk of HD may go through prior to testing and illustrated how favourable results may impact upon a person and their family. This could be of use to health professionals working in the area of genetic counselling.

Although the findings of the present study have provided a valuable contribution to the literature on the psychological impact of predictive genetic testing for HD, the limitations of the study should be acknowledged. It could be argued that the sample was biased because all participants were selected by a Consultant Clinical Geneticist who decided who to approach to take part in the study. Although potential participants could have been contacted through HD support groups or HD advisors, it was decided that it was more appropriate to recruit through the Consultant due to the sensitive nature of the research topic. She was able to use her clinical knowledge and experience to decide on the suitability of patients for inclusion in the study. Despite the researcher not having control over who was selected there was variation within the sample in terms of gender, age, marital status, length of time since favourable result, family experience

of HD, age when found out about risk of HD and coping abilities. There was also limited opportunity to be selective about participants given that the overall size of the population of interest was relatively small. It was therefore necessary to accept participants who expressed an interest in taking part and who met the inclusion criteria.

It would have been preferable to validate the findings of the study by asking some of the participants for their thoughts on the substantive theory and whether the categories had meaning for them. Unfortunately this was not possible due to limited time. However, the participants will be sent a copy of the findings and they will be invited to provide feedback. The findings were, however, validated through regularly going back to the raw data to check out that the theory was grounded in the participants' experiences, consulting the literature and keeping an audit trail which enabled the researcher to refer to memos and track the development of codes and categories and also decisions that were made during the analysis process. Discussions were also held with the researcher's supervisor about developing categories and their relationships to each other.

Another possible limitation was that all of the participants were at least 5 years post-test result with some of them interviewed as long as 10 years after the result. If they had been interviewed closer to the time of the test result then their perspective on the situation might have been quite different. Reflecting back and trying to remember how one felt many years ago could result in participants not providing an accurate account of their experiences at that time. On the other hand the participants have had the opportunity to think about genetic testing and consolidate its impact on their lives over a

longer period of time. This could have resulted in a richer and more reflective description of their experiences. For example, a participant commented in the study that at the time of testing and shortly afterwards she felt happy with her result, but now looking back she believes it was a false happiness.

4.7 Implications for Clinical Practice

The findings of this study have a number of important implications for clinical practice. Advances in medical science mean that more people are going to have the opportunity to have access to information about their future health status in terms of their genetic risk for various diseases. With this in mind there is the potential for people to struggle with the psychological impact of knowing they have a high chance of developing a serious illness or terminal disease (Shiloh, 1996). Furthermore, the results of predictive testing can impact negatively on the individual and their family. Shiloh (1996) highlighted the need for clinical psychologists to work in this area to assist both patients and health professionals in terms of being part of the genetic counselling team and becoming involved in more complex cases where there is a breakdown in family relationships at any stage of the genetic testing process. Individual or family therapy can be offered to patients, and clinical psychologists can also work on a consultancy basis with genetic counselling teams and provide supervision, advice and support in difficult cases (Shiloh, 1996). Clinical psychologists can also provide a role in the training of genetic counsellors on the psychological issues affecting people at risk of HD and the psychological consequences of genetic testing (Shiloh, 1996). In order to have the knowledge to provide this service there is a need for clinical psychology

training programs to incorporate genetic testing into their syllabus (Lerman, *et al.* 2000).

More specifically in relation to the findings of the present study, there is a need for clinical psychologists working in the area of predictive testing for HD to be aware that people who receive favourable test results may need long-term support for themselves and their families. For some participants it was a struggle to manage the uncertainty associated with living at risk of HD and psychological therapy at this stage may help to lessen depression and anxiety about the future. In addition family therapy may encourage a more open approach to testing and an understanding between family members as to why people want the test and how they would all feel about favourable or unfavourable results.

The results in this study suggest that people who have family members with HD may find it more difficult to cope with their favourable result. Therefore, people could be identified as being of higher risk of psychological difficulties if they receive the only favourable result amongst a family where HD is prevalent. Psychological support could be offered following the test result and again in the future in anticipation of the difficulties of seeing loved ones deteriorate as the disease progresses. Although many people who receive favourable or unfavourable results do not experience psychological difficulties and are able to move on with their lives, there are still those who struggle to adjust to their new genetic status. Clinical psychologists need to be mindful that each

case is different and that they may need to adapt their approach to suit the particular problems of the individual and their family.

4.8 Recommendations for Future Research

Through carrying out the present study a number of areas for further research have been identified. It was clear during the course of the research that HD is very much a family disease and that a test result not only impacts on the individual but also on the spouse, children, parents, siblings and cousins. More research studies which include the perspective of a variety of different family members on living at risk of HD, the genetic testing process and the test results would provide valuable information about HD in the context of a family system. There do not appear to be any research studies which focus exclusively on sibling relationships before, during or after the predictive testing process in terms of the impact on the relationship when some siblings decide to go for testing, how the test results are communicated to siblings and how the relationship is affected thereafter depending on the test result.

In the present study it was the sibling relationship which participants talked about the most in terms of how difficult it was to see a brother or sister deteriorate after they had received a favourable result. It would be interesting to find out how it feels to receive an unfavourable result amongst siblings who find out they don't have the HD gene and vice versa. A longitudinal study would be valuable looking at the sibling relationship at different time points such as before the test and then at various stages after the results particularly in terms of how the relationship changes as the sibling with HD deteriorates.

More research into the spouses or partners of people who are at risk of HD is needed in order to find out how they cope with the psychological journey from finding out about the risk through to life after the result. The literature and the findings from this study suggest that for some people who receive favourable results their marriages breakdown but this appears to be more related to the long-term stress of living in a family at risk of HD than the test result itself. It would be valuable to find out the spouses perspective at all stages of the journey, and whether they think life should move forward without difficulty after a favourable result. The substantive model proposed in the present study could be explored in a larger sample of people who received favourable results to see if it is meaningful for them.

4.9 Personal Reflections

At the start of the research my knowledge and clinical experience of HD was minimal. I had conducted neuropsychological assessments with a few patients who were starting to show early signs of the disease but I had not seen any patients in the more advanced stages. Although I had some idea about the complexities of predictive testing for the disease through discussions with a Consultant Clinical Geneticist and reading the literature, it wasn't until I was immersed in data collection and analysis that I realised just how difficult life is for people who have HD in their family. During the research process, the writing of a reflective diary and memos about developing categories helped to bring clarity to my emerging ideas and think about how the research was impacting on me, as well as how I thought I was perceived by the participants. I was always very mindful of how difficult it must be speaking to a stranger about deeply personal and

upsetting life experiences and tried my best to put the participants at ease. A few participants commented that they felt relaxed in my company and that they did not feel intimidated by the situation. It was beneficial to have clinical and research supervisors to speak to about some of the interview experiences which had been more emotional or thought-provoking. The courage and resilience demonstrated by the participants was humbling and made me realise how difficult and complex life can be for some people.

4.10 Conclusions

Huntington's disease is a devastating illness which has wide ranging implications for those who have the HD gene and also their family. As soon as people find out they are at risk of the disease, life changes and the future appears more uncertain. The psychological impact of living at risk can be unbearable for some individuals who therefore decide to take the predictive test as soon as possible. For others the uncertainty gives hope that they may not have the HD gene and they prefer to live this way. Both favourable and unfavourable test results can negatively affect a person and the response to the test can depend on the genetic status of other family members. Living in a family in which a parent and siblings have HD can be emotionally challenging for a person with a favourable result, and feelings of guilt can occur as well as having a sense of duty towards family members with HD. It therefore can be difficult for people who receive favourable results to distance themselves from HD and move forward with their lives. Some people who received their favourable results years ago and who have lost their parent and sibling to HD still struggle to forget about HD and move forward and there was a sense that for many people HD will always be a part of them. There is need for clinical psychologists to provide psychological support in the

form of individual and family therapy to people at-risk of HD from when they first enquire about testing through to a number of years after their favourable result. This thesis will finish as it started with a quote from another family member in Sulaiman (2007) which illustrates how a person who received a favourable result feels living in a family with HD. Along with the findings presented in this thesis the quote challenges the assumption that receiving a favourable result can put your mind at ease and free you from the burden of HD:

“My family has HD and I have to live with it the best way I know how. Most of the time I do manage that and get on with life. But, the lucky one? I don’t think so. Never once have I considered myself to be the lucky one (p.45)”

References

- Adam, S., Wiggins, S. & Whyte, P., Bloch, M., Shokeir, M.H.K., Soltan, H., *et al.* (1993). Five year study of prenatal testing for Huntington's disease: demands, attitudes and psychological assessment. *Journal of Medical Genetics*, 30, 549-556.
- Almqvist, E.W., Brinkman, R.R., Wiggins, S. & Hayden, M.R. (2003). Psychological consequences and predictors of adverse events in the first 5 years after predictive testing for Huntington's disease. *Clinical Genetics*, 64, 300-309.
- Benjamin, C.M. & Lashwood, A. (2000). United Kingdom experience of presymptomatic testing of individuals at 25% risk for Huntington's disease. *Clinical Genetics*, 58, 41-49.
- Binedell, J., Soldan, J.R. & Harper, P.S. (1998a). Predictive testing for HD: I. predictors of uptake in South Wales. *Clinical Genetics*, 54(6), 477-488.
- Binedell, J., Soldan, J.R. & Harper, P.S. (1998b). Predictive testing for HD: II qualitative findings from a study of uptake in South Wales. *Clinical Genetics*, 54(6), 489-496.
- Broadstock, M., Michie, S. & Marteau, T. (2000). Psychological consequences of predictive genetic testing: a systematic review. *European Journal of Human Genetics*, 8, 731-738.
- Brouwer-DudokdeWit, A.C., Savenije, A., Zoetewij, M.W., Matt-Kievit, A., & Tibben, A. (2002). A hereditary disorder in the family and the family life cycle. Huntington disease as a paradigm. *Family Process*, 41(4), 677-692.
- Bryant, A. & Charmaz, K. (2010). Grounded theory in historical perspective: An epistemological account. In A Bryant & K Charmaz (Eds). *The sage handbook of grounded theory*. (pp.31-57). London: Sage Publications Ltd.
- Carter, B. & McGoldrick, M. (1989). Overview: the changing family life cycle: A framework for family therapy. In B. Carter & M. McGoldrick (Eds). *The changing family life cycle – A framework for family therapy*. (2nd edn., pp.3-28). Needham Heights, MA: Allyn and Bacon.

Chapman, E. (2002). The social and ethical implications of changing medical technologies: The views of people living with genetic conditions. *Journal of Health Psychology*, 7, 195-206.

Chapman, E. & Smith, J.A. (2002). Interpretive phenomenological analysis and the new genetics. *Journal of Health Psychology*, 7, 125-130.

Charmaz, K. (1999). Stories of suffering: subjective tales and research narratives. *Qualitative Health Research*, 9(3), 362-382.

Charmaz, K. (2000). Grounded theory: Objectivist and constructivist methods. In N.K. Denzin & Y.S. Lincoln (Eds). *Handbook of qualitative research* (2nd edn., pp509-535). Thousand Oaks: Sage Publications Ltd.

Charmaz, K. (2008a). *Constructing grounded theory: A practical guide through qualitative analysis*. London: Sage Publications Ltd.

Charmaz, K. (2008b). Grounded theory. In J.A. Smith (Ed.) *Qualitative psychology: A practical guide to research methods*. (pp. 81-110). London: Sage Publications Ltd.

Charmaz, K. (2009). Shifting the grounds: Constructivist grounded theory methods. In J.M. Morse, P.N. Stern, J. Corbin, B. Bowers, K. Charmaz & A. Clark. (Eds). *Developing grounded theory: The second generation*. (pp.127-147). Walnut Creek, CA: Left Coast Press Inc.

Codori, A.M., Hanson, R. & Brandt, J. (1994). Self-selection in predictive testing for HD. *American Journal of Medical Genetics*, 54, 167-173.

Codori, A.M., Slavney, P.R., Young, C., Miglioretti, D.L. & Brandt, J. (1997). Predictors of psychological adjustment to genetic testing for Huntington's disease. *Health Psychology*, 16(1), 36-50.

Creswell, J.W. (1998). *Qualitative inquiry and research design: choosing among five traditions*. Thousand Oaks: Sage Publications.

Dancyger, C., Smith, J.A., Jacobs, C., Wallace, M. & Michie, S. (2010). Comparing family members' motivations and attitudes towards genetic testing for hereditary breast and ovarian cancer: a qualitative analysis. *European Journal of Human Genetics*, 18, 1289-1295.

Dawson, S., Kristjanson, L.J., Toye, C.M. & Flett, P. (2004). Living with Huntington's disease: need for supportive care. *Nursing and Health Sciences*, 6, 123-130.

Decruyenaere, M., Evers-Kiebooms, G., Boogaerts, A., Cassiman, J.J., Cloostermans, T., Demyttenaere, K. *et al.* (1996). Prediction of psychological functioning one year after the predictive test for Huntington's disease and impact of the test result on reproductive decision making. *Journal of Medical Genetics*, 33, 737-743.

Decruyenaere, M., Evers-Kiebooms, G., Boogaerts, A., Demyttenaere, K., Dom, R. & Fryns, J.P. (2005). Partners of mutation-carriers for Huntington's disease: forgotten persons? *European Journal of Human Genetics*, 13, 1077-1085

Decruyenaere, M., Evers-Kiebooms, G. & Boogaerts, A., Phillipe, K., Demyttenaere, K., Vandenberghe, W. *et al.* (2007). The complexity of reproductive decision-making in asymptomatic carriers of the Huntington's mutation. *European Journal of Human Genetics*, 15, 453-462.

Draucker, C.B., Martsof, D.S., Ross, R. & Rusk, T.B. (2007). Theoretical sampling and category development in grounded theory. *Qualitative Health Research*, 17, 1137-1148.

Dudok de Wit, A.C., Duivenvoorden, H.J., Passchier, J., Niermeijer, F. & Tibben, A. (1998). Course of distress experienced by persons at risk for an autosomal dominant inheritable disorder participating in a predictive testing program: An exploratory study. *Psychosomatic Medicine*, 60, 543-549.

Duisterhof, M., Trijsburg, R.W., Niermeijer, M.F., Roos, R.A.C. & Tibben, A. (2001). Psychological studies in Huntington's disease: making up the balance. *Journal of Medical Genetics*, 38, 852-861.

Etchegary, H. (2006). Discovering the family history of Huntington's disease. *Journal of Genetic Counseling*, 15(2), 105-117.

Etchegary, H. (2010). I put it on the back burner most days: Living with chronic risk. *Health (London)*, 22nd December 2010, 1-17.

Evers-Kiebooms, G. & Decruyenaere, M. (1998). Predictive testing for Huntington's disease: a challenge for persons at risk and for professionals. *Patient Education and Counselling*, 35(1), 15-26.

Evers-Kiebooms, G., Nys, K., Harper, P., Zoetewij, M., Durr, A., Jacopini, G. *et al.* (2002). Predictive DNA-testing for Huntington's disease and reproductive decision

making: a European collaborative study. *European Journal of Human Genetics*, 10, 167-176.

Evers-Kiebooms, G., Swerts, A., Cassiman, J.J & Van den Berghe, H. (1989). The motivation of at-risk individuals and their partners in deciding for or against predictive testing for Huntington's disease. *Clinical Genetics*, 35, 29-40.

Falk, K., Granger, B.B., Swedberg, K., & Ekman, I. (2007). Breaking the vicious circle of fatigue in patients with chronic heart failure. *Qualitative Health Research*, 17(8), 1020-1027.

Gargiulo, M., Lejeune, S. & Tanguy, M.L., Lahlou-Laforet, K., Faudet, A., Cohen, D. *et al.* (2009). Long-term outcome of presymptomatic testing in Huntington's disease. *European Journal of Human Genetics*, 17, 165-171.

Glaser, B.G. (1978). *Theoretical sensitivity*. Mill Valley CA: Sociology Press.

Glaser, B.G. (1992). *Basics of grounded theory analysis: Emergence versus forcing*. Mill Valley CA: Sociology Press.

Glaser, B.G., & Strauss, A.L. (1967) *The discovery of grounded theory: Strategies for qualitative research*. Chicago: Aldine.

Gusella, J.F., Wexler, N.S., Conneally, P.M. Naylor, S.R., Anderson, M.A., Tanzi, R.E. *et al.* (1983). A polymorphic DNA marker genetically linked to Huntington's disease. *Nature*, 306, 234-238.

Hamann, , H.A., Smith, T.W., Smith, H.R., Croyle, R.T., Ruiz, J.M., Kircher, J.C. & Botkin, J.R. (2008). Interpersonal responses among sibling dyads tested for BRCA1/BRCA2 Gene Mutations. *Health Psychology*, 27(1), 100-109.

Harper, P.S., Lim, C. & David, C. (2000). Ten years of presymptomatic testing for Huntington's disease: the experience of the UK Huntington's Disease Prediction Consortium. *Journal of Medical Genetics*, 37, 567-571.

Hayes, C.V. (1992). Genetic testing for Huntington's disease: a family issue. *New England Journal of Medicine*, 327, 1449-1451.

Holtslander, L.F. & Duggleby, W.D. (2009). The hope experience of older bereaved women who cared for a spouse with terminal cancer. *Qualitative Health Research*, 19(3), 388-400.

Huggins, M., Bloch, M., Wiggins, S., Adam, S., Suchowersky, O., Trew, M. *et al.* (1992). Predictive testing for Huntington disease in Canada: Adverse effects and unexpected results in those receiving a decreased risk. *American Journal of Medical Genetics*, 42, 508-515.

Huntington's Disease Association (2008). General information about Huntington's and the Huntington's disease association: Fact sheet 1. Retrieved 8 July 2009 from <http://www.hda.org.uk/download/fact-sheets/HD-About-The-HDA.pdf>

Huntington's Disease Association (2011). Predictive testing for Huntington's disease. Fact sheet. Retrieved 10 October 2011 from <http://www.hda.org.uk/download/fact-sheets/HD-Predictive-Testing.pdf>

Keenan, K.F., Miedzybrodzka, Z., van Teijlingen, E., McKee, L. & Simpson, S.A. (2007). Young people's experiences of growing up in a family affected by Huntington's disease. *Clinical Genetics*, 71, 120-129.

Kessler, S. (1988). Preselection: A family coping strategy in Huntington disease. *American Journal of Medical Genetics*, 31, 617-621.

Kessler, S. (1993a). Forgotten person in the Huntington disease family. *American Journal of Medical Genetics*, 48(3), 145-150.

Kessler, S. (1993b). The spouse in the Huntington's disease family. *Family Systems Medicine*, 11(2), 191-199.

Kessler, S. (1994). Predictive testing for Huntington's disease: a psychologist's view. *American Journal of Medical Genetics*, 54, 161-166.

Klitzman, R., Thorne, D., Williamson, J., Chung, W & Marder, K. (2007). Decision-making about reproductive choices among individuals at-risk for Huntington's disease. *Journal of Genetic Counselling*, 16(3), 347-362.

Larsson, M.U., Luszcz, M.A., Bui, T.H. & Robins Wahlin, T.B. (2006). Depression and suicidal ideation after predictive testing for Huntington's disease: A two-year follow up study. *Journal of Genetic Counselling*, 15(5), 361-374.

Lawson, K., Wiggins, S.G., Tiffinie, A.S., Bloch, M. & Hayden, M.R. (1996). Adverse psychological events occurring in the first year after predictive testing for Huntington's disease. *Journal of Medical Genetics*, 33(10), 856-862.

- Lerman, C., Croyle, R.T., Tercyak, K.P. & Hamann, H. (2002). Genetic testing: psychological aspects and implications. *Journal of Consulting and Clinical Psychology*, 70(3), 784-797.
- MacLeod, R., Craufurd, D., and Booth, K. (2002). Patients' perceptions of what makes genetic counselling effective: an interpretive phenomenological analysis. *Journal of Health Psychology*, 7(2), 145-156.
- McAllister, M. (2001). Grounded theory in genetic counseling research. *Journal of Genetic Counseling*, 10(3), 233-250.
- Meiser, B. & Dunn, S. (2000). Psychological impact of genetic testing for Huntington's disease: an update of the literature. *Journal of Neurology, Neurosurgery and Psychiatry*, 69, 574-578.
- Morse, J.M. (2001). Situating grounded theory within qualitative inquiry. In R.S. Schreiber & P.S. & Stern (Eds.) *Using grounded theory in nursing*. (pp.1-16). New York: Springer Publishing Company.
- Nunkoosing, K. (2005). The problems with interviews. *Qualitative Health Research*, 15(5), 698-706.
- Quaid, K.A., Sims, S.L., Swenson, M.M., Harrison, J.M. Moskowitz, C., Stepanov, N. *et al.* (2008). Living at risk: Concealing risk and preserving hope in huntington's disease. *Journal of Genetic Counselling*, 17, 117-128.
- Quaid, K.A. & Wesson, M.K. (1995). Exploration of the effects of predictive testing for Huntington disease on intimate relationships. *American Journal of Medical Genetics*, 57, 46-51.
- Quarrell, O. (2008). *Huntington's disease. The facts*. Oxford: Oxford University Press.
- Rawlins, M. (2010). Huntington's disease out of the closet? *The Lancet*, 376(9750), 1372-1373.
- Richards, F. (2004). Couples' experiences of predictive testing and living with the risk or reality of Huntington disease: A qualitative study. *American Journal of Medical Genetics*, 126(A), 170-182.

Richards, F.H. & Rea, G. (2005). Reproductive decision-making before and after predictive testing for Huntington's disease: An Australian perspective. *Clinical Genetics*, 67, 404-411.

Schreiber, R.S. (2001). The "how to" of grounded theory: Avoiding the pitfalls. In R.S. Schreiber & P.N. Stern. (Eds). *Using grounded theory in nursing* (pp.55-83). New York: Springer Publishing Company.

Shiloh, S. (1996). Genetic counseling: A developing area of interest for psychologists. *Professional Psychology: Research and Practice*, 27(5), 475-486.

Smith, J.A., Michie, S., Stephenson, M. & Quarrell, O. (2002). Risk perception and decision-making processes in candidates for genetic testing for Huntington's Disease. An interpretive phenomenological analysis. *Journal of Health Psychology*, 7(2), 131-144.

Smith, J.A. & Osborn, M. (2008). Interpretative phenomenological analysis. In J.A. Smith (Ed.) *Qualitative psychology: A practical guide to research methods*. (pp.53-80). London: Sage Publications Ltd.

Sobel, S.K. & Cowan, D.B. (2000a). The process of family reconstruction after DNA testing for Huntington's disease. *Journal of Genetic Counselling*, 9(3), 237-251.

Sobel, S.K. & Cowan, D.B. (2000b). Impact of genetic testing for Huntington's disease on the family system. *American Journal of Medical Genetics*, 90, 49-59.

Starks, H. and Trinidad, S.B. (2007). Choose your method: A comparison of phenomenology, discourse analysis and grounded theory. *Qualitative Health Research*, 17(10), 1372-1380.

Strauss, A.M., & Corbin, J. (1998) *Basics of qualitative research: techniques and procedures for developing grounded theory* (2nd edn). California: Sage Publications

Sulaiman, S. (2007). *Learning to live with Huntington's disease: One family's story*. London: Jessica Kingsley Publishers

The Huntington's Disease Collaborative Research Group (1993). A novel gene containing the trinucleotide repeat that is expanded and unstable on Huntington's Disease chromosomes. *Cell*, 72, 971-983.

Tibben, A. (2007). Predictive testing for Huntington's disease. *Brain Research Bulletin*, 72, 165-171.

Tibben, A., Duivenvoorden, H.J., Niermeijer, M.F., Vegter-Van der Vlis, M., Roos, R.A.C. & Verhage, F. (1994). Psychological Effects of Presymptomatic DNA Testing for Huntington's Disease in the Dutch Program. *Psychosomatic Medicine*, 56, 526-532.

Tibben, A., Frets, P.G., van de Kamp, J.J.P., Niermeijer, M.F., Vegter-van der Vlis, M., Roos, R.A.C., Rooymans, H.G.B. *et al.* (1993b). On attitudes and appreciation 6 months after predictive DNA testing for Huntington disease in the Dutch program. *American Journal of Medical Genetics*, 48, 103-111.

Tibben, A., Frets, P.G., van den Kamp, J.J.P., Niermeijer, M.F., Vegter-van der Vlis, M., Roos, R.A.C., van Ommen, G-J.B., *et al.* (1993a). Presymptomatic DNA testing for Huntington's Disease: Pretest attitudes and expectations of applicants and their partners in the Dutch program. *American Journal of Medical Genetics*, 48, 10-16.

Tibben, A., Tinman, R., Bannick, E.C. & Duivenvoorden, H.J. (1997). Three-year follow-up after presymptomatic testing for HD in tested individuals and partners. *Health Psychology*, 16, 20-35.

Timman, R., Roos, R., Matt-Kievit, A. & Tibben, A. (2004a). Adverse effects of predictive testing for Huntington Disease underestimated: Long-term effects 7-10 years after the test. *Health Psychology*, 23(2), 189-197.

Timman, R., Stijen, T. & Tibben, A. (2004b). Methodology in longitudinal studies on psychological effects of predictive DNA testing: A review. *Journal of Medical Genetics*, 41, e100. BMJ Group. Retrieved from <http://jmg.bmj.com/cgi/doi/10.1136/jmg.2003.017541>

Van Riper, M. (2005). Genetic testing and the family. *Journal of Midwifery and Women's Health*, 50(3), 227-223.

Van't Spijker, A. & ten Kroode, H.F.J. (1997). Psychological aspects of genetic counselling: A review of the experience with Huntington's disease. *Patient Education and Counselling*, 32, 33-40.

Walker, D. and Myrick, F. (2006). Grounded theory: an exploration of process and procedure. *Qualitative Health Research*, 16(4), 547-559.

Wiggins, S., Whyte, P., Huggins, M., Adam, A., Theilmann, J., Bloch, M. *et al.* (1992). The psychological consequences of predictive testing for Huntington's disease. *New England Journal of Medicine*, 327, 401-405.

Williams, J.K., Schutte, D.L., Evers, C. & Holkup, P.A. (2000a). Redefinition: Coping with normal results from predictive gene testing for neurodegenerative disorders. *Research in Nursing and Health*, 23, 260-269.

Williams, J.K., Schutte, D.L., Holkup, P.A., Evers, C. & Muilenberg, A. (2000b). Psychosocial impact of predictive testing for Huntington's disease on support persons. *American Journal of Medical Genetics*, 96(3), 353-359.

Willig, C. (2008). *Introducing qualitative research in psychology*. Maidenhead: Open University Press.

Yardley, L. (2000). Dilemmas in qualitative health research. *Psychology and Health*, 15, 215-228.

Yardley, L. (2008). Demonstrating validity in qualitative psychology. In J.A. Smith (Ed.) *Qualitative psychology: A practical guide to research methods*. (pp. 235-251). London: Sage Publications Ltd.

Participant Information Sheet

Study title

Favourable results from predictive testing for Huntington's disease: An exploration of the long-term impact on close relationships.

Invitation Paragraph

You are being invited to take part in a research study. Before you decide whether to take part it is important for you to understand why the research is being done and what it will involve. Please take time to read this information carefully and discuss it with others if you wish. Please ask us if there is anything which is not clear or if you would like more information. Take time to decide whether you wish to take part. Thank you for reading this.

What is the purpose of this study?

As part of my Doctoral training in Clinical Psychology I am carrying out a study that involves exploring the experiences of people who received favourable results from predictive genetic testing for Huntington's disease at least five years ago. Participation in this research study does not require you to undertake any medical tests or provide blood samples.

I am interested in exploring the impact of a favourable result on your life in general and in particular on your close relationships, for example with a spouse/partner, parents, siblings and friends. This study aims to record your thoughts and feelings about receiving your favourable result. I am not medically trained so therefore am unable to answer any questions you may have about your health status. However I can provide contact details of health professionals who can discuss with you any concerns you may have about your health status. The results of the study should provide useful information about the impact of receiving favourable results from genetic testing for Huntington's disease.

The total duration of the study is one interview lasting approximately one hour with the possibility of one further interview lasting between 40 and 60 minutes.

Why have I been chosen?

You are being asked to participate because you received a favourable result from predictive genetic testing for Huntington's disease at least five years ago. I am interested in finding out how this result has impacted on your life and your close relationships.

Do I have to take part?

It is up to you to decide whether to take part. If you do decide to take part, you will be asked to sign a consent form. You are still free to withdraw from the study at any time and without giving a reason. A decision to withdraw at any time, or a decision not to take part, will not affect the standard of care you receive.

What will happen to me if I take part?

You will be asked to attend an appointment at the Department of Clinical Neuropsychology, Aberdeen Royal Infirmary, Aberdeen. Alternatively, if this is inconvenient for you it might be possible to meet with you in your own home. The appointment will involve an interview lasting approximately one hour in which you will be asked to describe your experiences of receiving a favourable result from genetic testing for Huntington's disease. You do not have to answer any questions which you do not wish to answer. The interview will be tape-recorded and the tape will be destroyed as soon as the interview has been transcribed. At the end of the interview I will ask if you would mind being contacted again if I have further questions which I would like to ask you.

What are the possible disadvantages and risks of taking part?

There are no known risks associated with taking part in this research study. Sometimes people can feel upset when talking about important or stressful events in their lives. If during the interview you feel upset, you are free to stop at any time, without providing a reason, and you can make the decision if you wish to continue. If you feel that you would like to talk further about any issues that were upsetting for you in the interview, support will be available to you from Dr Simpson, Consultant in Clinical Genetics.

What are the possible benefits of taking part?

There are no known immediate benefits to taking part in the research study. It is hoped that this study will provide useful information relating to the long-term issues and the support needs affecting individuals who receive favourable results from predictive testing for HD.

What if something goes wrong?

If you wish to complain, or have any concerns about any aspect of the way you have been approached or treated during this study, the usual National Health Service complaints mechanisms will be available to you.

Will my taking part in this study be kept confidential?

Information which is collected about you will be kept strictly confidential. Any information which leaves the hospital will have your name and address removed so that you cannot be recognised from it.

Personal data collected during the study will be destroyed after the study is finished.

During or after the interview I have cause for concern for the safety or well-being of you or others, you need to be aware that in your own best interests, and after consulting with you, an appropriate person such as a healthcare professional will be informed of my concerns.

What will happen to the results of the study?

The results from this study will be written up and submitted for academic review as part of my qualifications as a Trainee Clinical Psychologist on the University of Edinburgh Clinical Psychology Doctoral course. Your information will be anonymous and your privacy will be respected. Your name will not be used in any reports as code numbers will be used instead.

I will send you a summary of the main results of the study if you would like to receive this. Presentations will also be used so that practitioners who work with people who are at risk of HD who are receiving genetic testing for HD can hear about the results of the study. No participants will be identified in any publication or presentation.

Who is organising and funding the research?

This research is jointly organised by NHS Grampian and the University of Edinburgh.

Who has reviewed the study?

This study has been reviewed and approved by the North of Scotland Research Ethics Committee and the University of Edinburgh Clinical Psychology Ethics Committee.

Contact for further information

If you require any further information please do not hesitate to contact me:

Dianne Beastall, at Department of Clinical Neuropsychology, Ward 40, Aberdeen Royal Infirmary, Aberdeen, AB25 2NZ

or contact by telephone on 01224 556147 (9-5pm)

or by email on dianne.beastall@nhs.net

You can also contact Dr Emma Hepburn, Clinical Psychologist, Research Supervisor, at Neuropsychology Department, Ward 40, Aberdeen Royal Infirmary, Aberdeen, AB25 2NZ.

or contact by telephone on 01224 554350.

Thank you for considering taking part in this study.



Dr Anne Swends
Cranfield Health Psychology
Department of Health Psychology
200-140
Preston Road, Bury St Edmunds
Suffolk IP8 3PH
Telephone 01204 254747 (B-4425)

First Name: _____

Signature: _____

CONSENT FORM

Dr Anne Swends, Cranfield Health Psychology, is conducting a research project to explore the experiences of people who have been in a close relationship with someone who has a mental health problem.

Dr Anne Swends

I confirm that I have read and understood the information sheet and have had the opportunity to discuss the information with me and have had time to think about it. I have signed this form and have had time to think about it. ☐

Appendix 2: Consent Form

I understand that my participation is voluntary and that I can withdraw at any time without giving any reason. ☐

I agree to have the research information stored on a secure computer and to be contacted by email. ☐

I agree to take part in the study. ☐

Name of Participant: _____

Signature: _____

Date: _____



Dianne Beastall
Trainee Clinical Psychologist
Department of Neuropsychology
Ward 40
Aberdeen Royal Infirmary
Foresterhill
Aberdeen AB25 2ZN
Telephone 01224 556147 (9-5pm)

E-mail dianne.beastall@nhs.net

Study Number:

CONSENT FORM

Title of Project: Favourable results from predictive testing for Huntington's disease: An exploration of the long-term impact on close relationships.

Name of Researcher: Dianne Beastall

I confirm that I have read and understand the information sheet dated (version) for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily.

Please initial box

☐

I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

☐

I agree to have the research interview tape-recorded on condition that the tape is destroyed once the interview has been transcribed.

☐

I agree to take part in the above study.

☐

Signature of Patient

Date

Signature

Signature of Researcher

Date

Signature

Would you like to receive a summary of the research findings at the end of the study:

Please Circle
YES / NO

Appendix 3:

Approval from Research Ethics Committee of the University of Edinburgh Doctorate in Clinical Psychology

Quota status: 218.08MB / 1024.00MB (21.30%)

Search Results: Re: NHS Ethics (27 of 61)

as: Move | Copy This message to Back to Search Results
te | Reply | Reply to All | Forward | Redirect | Blacklist | Whitelist | Message Source | Save as | Print |
ort as Spam | Report as Innocent

Date: Mon, 16 Feb 2009 14:58:09 +0000 [16/02/2009 14:58:09 BST]

From: Ethel Quayle <equayle@staffmail.ed.ac.uk>

To: DJ Beastall <D.J.Beastall@sms.ed.ac.uk>, Evelyn Kelly <Evelyn.A.Kelly@ed.ac.uk>

Subject: Re: NHS Ethics

ers: Show All Headers

Dianne,
r submission was discussed at today's Panel. The following comments were
e:

Panel thought that this was an interesting and well-prepared proposal. No
nges are
quired with regard to research or ethical issues. It was felt that when
mitting to
NHS Ethics Committee it would be necessary to actually specify what some of
ical issues are, rather than simply generating ways of addressing them.

you are pleased with this. Let me know how the work is going,

t wishes,

el

Beastall wrote:

[Quoted Text]

Ethel

st to let you know that unfortunately I have been unable to book my NHS
thics
plication into the next meeting (March 12th) as it is fully booked. It is
t a major
t-back as I go on holiday for 2 weeks on 18th March so wouldnt have been
le to do
ything anyway. My application is now booked in for the meeting on 26th
rch and it
s a deadline for submission on 5th march. It is maybe for the best as this
ves me
re time to hear back from course ethics next week and deal with the comments
ey
ise, get their letter, sponsorship letter and your signature. Sheila
rently has a

North of Scotland Research Ethics Committee
South of Scotland
East of Scotland
West of Scotland
Aberdeen



Telephone: 01224 255555
Fax: 01224 255556
Email: nres@nhs.uk

15 April 2020

Dr Charles Edwards
Manager, Dept of Psychology
Clinical Psychology Dept
Glasgow Building
Royal Cornhill House
ABERDEEN
AB9 8QH

Dear Dr Edwards

Appendix 4:

Approval from North of Scotland Research Ethics Service

REG reference number:

Title of study:

Confirmation of ethical advice:

On behalf of the Committee, I am pleased to inform you that your application for the above research has been approved by the North of Scotland Research Ethics Service.

Details of the research:

The Committee has considered your application and is satisfied that the research is in the interests of the community and that the research is of a high standard. The Committee has also considered the ethical aspects of the research and is satisfied that the research is ethically sound.

Details of the research:

The research is of a high standard and is in the interests of the community.

The research is of a high standard and is in the interests of the community.

The research is of a high standard and is in the interests of the community.

North of Scotland Research Ethics Committees

Summerfield House
2 Eday Road
Aberdeen
AB15 6RE

Telephone: 01224 558474
Facsimile: 01224 558609
Email: nosres@nhs.net



21 April 2009

Dr Dianne Beastall
Trainee Clinical Psychologist
Clinical Psychology Dept
Clerkseat Building
Royal Cornhill Hospital
ABERDEEN
AB25 2ZH

Dear Dr Beastall

Full title of study: Favourable results from predictive testing for
Huntington's disease: An exploration of the long-term
impact on close relationships.

REC reference number: 09/S0801/39

Thank you for your letter of 17 April 2009, responding to the Committee's request for further information on the above research and submitting revised documentation.

Confirmation of ethical opinion

On behalf of the Committee, I am pleased to confirm a favourable ethical opinion for the above research on the basis described in the application form, protocol and supporting documentation as revised, subject to the conditions specified below.

Ethical review of research sites

The Committee has not yet been notified of the outcome of any site-specific assessment (SSA) for the research site(s) taking part in this study. The favourable opinion does not therefore apply to any site at present. We will write to you again as soon as one Research Ethics Committee has notified the outcome of a SSA. In the meantime no study procedures should be initiated at sites requiring SSA.

Conditions of the favourable opinion

The favourable opinion is subject to the following conditions being met prior to the start of the study.

Management permission or approval must be obtained from each host organisation prior to the start of the study at the site concerned.

Management permission at NHS sites ("R&D approval") should be obtained from the relevant care organisation(s) in accordance with NHS research governance arrangements.

Guidance on applying for NHS permission is available in the Integrated Research Application System or at <http://www.rdforum.nhs.uk>.

Approved documents

The final list of documents reviewed and approved by the Committee is as follows:

Document	Version	Date
Contact Sheet	1	27 February 2009
Recruitment Letter	1	27 February 2009
Ethel Quayle - CV		
Participant Consent Form	1	27 February 2009
Letter of invitation to participant	1	27 February 2009
Interview Schedules/Topic Guides	1	27 February 2009
Peer Review		02 March 2009
Letter from Sponsor		17 February 2009
Protocol	1	27 February 2009
Investigator CV		27 February 2009
Application	2.0	27 February 2009
Response to Request for Further Information	N/A	17 April 2009
Participant Information Sheet	2	08 April 2009
Covering Letter	N/A	08 April 2009

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

After ethical review

Now that you have completed the application process please visit the National Research Ethics Website > After Review

You are invited to give your view of the service that you have received from the National Research Ethics Service and the application procedure. If you wish to make your views known please use the feedback form available on the website.

The attached document “After ethical review –guidance for researchers” gives detailed guidance on reporting requirements for studies with a favourable opinion, including:

- Notifying substantial amendments
- Progress and safety reports
- Notifying the end of the study

The NRES website also provides guidance on these topics, which is updated in the light of changes in reporting requirements or procedures.

We would also like to inform you that we consult regularly with stakeholders to improve our service. If you would like to join our Reference Group please email referencegroup@nres.npsa.nhs.uk.

With the Committee's best wishes for the success of this project

Yours sincerely

Dr Angus J Thompson
Chair

Dr Denise Donatelli

Trainee Clinical Psychologist

Department of Clinical

Neuropsychology

Warr 40

Aberdeen Royal Infirmary

Foresterhill

Aberdeen

AB25 2ZN

Date: 05/05/2018

Our Ref: 2008PC005

Forwards to:

Email: 31421

Direct Line: 01224 334121

Dear Dr Donatelli

I am writing to you

in relation to

your request for

a review of your

application for

a review of your

application for

a review of your

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Appendix 5:

Management Approval from NHS Grampian R&D

Thank you very much for the copy of your application. I am pleased to confirm that your project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:

All resources will be in place for the project. The project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:

It is particularly important that you inform us of any changes to the study protocol.

The R&D Office has the following information on the project. The project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:

- The project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:
- The project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:
- The project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:
- The project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:
- The project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:

Please inform us of any changes to the project. The project is now registered with the R&D Office for Health and Care Research Office. The project has R&D Management Approval for a period of 12 months from 01/05/2018 to 01/05/2019. This is based on the documents received from your project. The following approvals are in place:

Dr Dianne Beastall
Trainee Clinical Psychologist
Department of Clinical
Neuropsychology
Ward 40
Aberdeen Royal Infirmary
Foresterhill
Aberdeen
AB25 2ZN

Date 05/05/2009
Our Ref 2009PC005

Enquiries to
Extension 51121
Direct Line 01224 551121

Dear Dr Beastall

Management Approval for Non-Commercial Research

NOSRES Ref: 09/S0801/039
Project title: Favourable results from predictive testing for Huntington's disease: an exploration of the long-term impact on close relationships

Thank you very much for sending all relevant documentation. I am pleased to confirm that the above project is now registered with the NHS Grampian Research & Development Office. The project has R & D Management Approval to proceed locally from 08/05/2009 to 07/03/2010. This is based on the documents received from yourself and the relevant Approvals being in place.

All research with an NHS element is subject to the Research Governance Framework for Health and Community Care (2006, 2nd edition), and as Chief or Principal Investigator you should be fully committed to your responsibilities associated with this.

It is particularly important that you inform us when the study terminates.

The R&D Office must be notified immediately and any relevant documents forwarded to us if any of the following occur:

- A change of Principal Investigator, Chief Investigator or any additional research personnel
- Premature project termination
- Any amendments – substantial or non-substantial (particularly a study extension)
- Any change to funding or any additional funding
- Any Serious Adverse Events

Please also forward any documents relating to any of the above to the R&D Office.

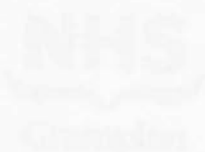
We hope the project goes well, and if you need any help or advice relating to your R&D Management Approval, please do not hesitate to contact the office.

Yours sincerely

Ms Pat Duff
Research and Development Manager

Dear Sir/Madam

Consultant
Dr Shafiq A. Hameed
North of Scotland Regional Genetics
Service
Forth House, Argyll House, Central
Road
Aberdeen AB25 2ZB
Telephone: 01224 572120
Fax: 01224 554190
E-mail: shafiq.hameed@nhs.uk



Dear

Enclosed is a letter from Professor Yehuda in Jerusalem's Hebrew University
Exploration of the Long-term Impact on Child Neurodevelopment

I write to invite you to take part in a research study. Myself and I have been selected a
possible participant for the study. The study is a research project to investigate the
neurodevelopmental effects of the long-term impact on child neurodevelopment. The
study will be conducted in the form of a questionnaire and a follow-up visit.
Details for the study are as follows: If you wish to participate, please contact the study team.

Appendix 6:

Letter of Invitation

If, after reading the letter, you wish to participate in the study, please contact the study team, or
write to the study team at the following address: Dr Shafiq A. Hameed, Consultant Geneticist, North of Scotland
Regional Genetics Service, Forth House, Argyll House, Central Road, Aberdeen AB25 2ZB. The study team will
then get in touch with you to arrange the study.

If you have any questions, please contact the study team.

Yours sincerely

Dr Shafiq A. Hameed
Consultant Geneticist

NHS Grampian

Consultant:
Dr Sheila A Simpson
North of Scotland Regional Genetics
Service
First Floor Argyll House Cornhill
Road
Aberdeen AB25 2ZR
Secretary Direct Line: 01224 552120
Fax: 01224 559390
Inverness 01463 705902



Dear

Favourable Results from Predictive Testing in Huntington's Disease: An Exploration of the Long—term Impact on Close Relationships.

I write to invite you to take part in a research study. More than 5 years ago you received a favourable presymptomatic predictive test result for Huntington's disease, and this research study aims to investigate outcomes for you and others in your position. The enclosed information sheet explains about the research project and includes contact details for the researchers if you wish to contact them to find out more about this study.

If, after reading the Information Sheet you decide you are interested in taking part, or wish to hear more about the study, please complete the enclosed Contact Sheet and return it to the researcher using the stamped, addressed envelope provided. The researcher will then get in touch with you to arrange further contact.

If you have any questions please do not hesitate to contact me.

Yours sincerely

Dr Sheila A Simpson
Consultant in Clinical Genetics

INTERVIEW GUIDE

Charmaz (2008) provides a list of sample grounded theory interview questions about a life change. In terms of the present study the questions used in the interviews might include:

Sample initial open-ended questions

- * "Tell me about happened when you found out the results of the genetic testing for HD?",
- * "What was going on in your life then? How would you describe how you viewed your life and relationships before the test result? How, if at all has your view of life changed?"

Sample intermediate questions

- * "Tell me about your thoughts and feelings when you learned about your test result?"
- * "Who, if anyone was involved? When was that? How were they involved?"
- * "Tell me how you learned to cope with the test result?"
- * "How did your family/spouse/partner react to your test result?"
- * "What positive changes have occurred in your life since receiving the favourable result?"
- * "What negative changes, if any, have occurred in your life since receiving the favourable result?"
- * "Tell me about how you would describe the person you are now."
- * "Could you describe the most important lessons you learned through receiving a favourable result from genetic testing?"
- * "Who has been the most helpful to you during this time? How has she/he been helpful?"
- * "In what ways (if any) are your present relationships affected by your favourable test result?"

Sample ending questions

- * "Tell me about your strengths that you discovered or developed through adapting to your test result?"
- * "After having these experiences, what advice would you give to someone who has just discovered that he/she does not have the HD gene?"
- * "Is there anything else that you think I should know to understand your experiences better?"